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**INFORMED CONSENT FOR
 GENETIC TESTING**
 In compliance with the New York
 State Civil Law: Section 79-L

CLIA ID: 11D0703390 GA License: 044-146 MD License: 1241 FL License: 800025664 NY PFI: 8159

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.)

1. Test Description (General): Instructions to Genetic Health Provider: Please check those that apply

- DNA (Molecular) PCR Amplification for mutation detection.
- Organic Acids Gas chromatographic mass spectrometric separation to identify urine organic acids
- Neurochemicals HPLC separation with electrochemical and/or UV and/or fluorescence detection of CSF metabolites to identify possible inherited metabolic diseases of central nervous system.
- Acylcarnitine In-born errors of fatty acid and amino acid metabolism may be identified by tandem MS/MS.
- Carnitine In-born errors in Carnitine metabolism may be identified by tandem MS/MS.
- Amino Acids Inborn errors of amino acid metabolism may be identified by HPLC/spectrophotometry
- Pyruvate/Lactate Nonspecific markers for possible metabolic disease by Enzymology/spectrophotometry
- Enzymology Specific enzyme activity measurement for the detection of various inherited diseases using various methodologies

2. Purpose of the test: The potential benefit of this study is to obtain the possible diagnosis of an inherited disease. It may also determine which family members may be carriers of the trait.

3. 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. A negative result may/may not rule out possible diagnosis depending on clinical history and quality/type of specimen tested. The individual may wish to consider further independent testing, consult a personal physician or pursue genetic counseling.

4. General description of diseases or conditions (symptoms may include):

Movement disorder	Developmental delay	Seizures	Metabolic acidosis
Encephalopathy	Failure to thrive	Hypoglycemia	Hyperammonemia
Skeletal myopathy	Sudden unexplained death	Cardiomyopathy	Ketosis
Mitochondrial disease	Other (as specified by ordering MD) _____		

5. Level of certainty: Is test-specific and determined by the methods employed, patient's clinical history and sometimes by the nature of the patient's condition at time of sampling. There is always a small possibility of an error or failure in sample analysis; this is true with complex testing in any laboratory.

6. Disclosing Test Results: The following are the categories of persons or organizations that test results may be released to. These include, but are not limited to: Hospitals or laboratories involved in the patient's care, referring physician(s) and primary care providers, other physician groups (consultants, surgeons), insurance companies (as provided by patient or the referring physician for payment purposes), and other professionals involved in patient care that assist Medical Neurogenetics in carrying out treatment, payment, and operational activities. Results are kept confidential. Medical Neurogenetics complies with security and privacy statutes of the federal Health Information Portability and Accountability Act (HIPAA). If patient chooses to specifically declare where results may be released (other than the referring institution and ordering physician), please provide these *in writing* to the Compliance Officer, Medical Neurogenetics.

7. Test Authorization:

- a. No clinical tests other than those authorized shall be performed on the sample.
- b. Remaining LABELED patient sample may be held for 10 years for further diagnostic or confirmatory testing after which time the sample may be discarded. A portion of the sample may be made anonymous (name and identifiers removed) and used for research purposes. Any results obtained could not be related to the original source so no results would be reported.
- c. The patient (guardian) may restrict or withdraw consent, *in writing*, to Medical Neurogenetics' Compliance Officer, to exclude the patient specimen(s) from being used for research, except to the extent that Medical Neurogenetics has already used them in reliance upon prior consent.

8. This written consent is signed by the person who is the subject of the test (or if that person lacks capacity to consent or is a minor, signed by the person authorized to consent for that person.) This form is kept on file at the referring facility. A copy may or may not be available at Medical Neurogenetics.

_____, Or _____
 Signature of Patient Date

 Authorized Signature Relationship

 Name of Patient (Please Print Clearly) Name of Ordering MD (Please Print Clearly)

 Referring Facility (Please Print Clearly) Date Specimen Collected/Sent