NewYork-Presbyterian
The University Hospital of Columbia and Cornell

45350

***HIGH READING LEVEL**
INFORMED CONSENT FORM FOR AMINO ACID PROFILES

IF NO PLATE, PRINT NAME, SEX AND MEDICAL RECORD NO.

Type of Specimen: Plasma in Sodium/Lithium Heparin tube, 1-2 ml.
Delivered to Laboratory **immediately** after drawing.
Please contact the Biochemical Genetics Laboratory with specific questions: 212-305-6248

This form must be completely filled out, signed by patient, parent/legal guardian or legal next of kin and maintained in the patient's Medical Record at the physician's office. Additionally, a copy of this consent must be included with the specimen when received in the laboratory.

ICD-9 Code: _____

Requesting Physician: _____

Phone: _____

Pager #: _____

Office Fax: _____

Reason for Testing: _____

Date Specimen Collected (month/date/year): _____

TO THE PATIENT/PARENT/LEGAL GUARDIAN:
Please read the following carefully and discuss with your ordering physician/person obtaining consent before signing consent.

_____ has explained to me, in a way that I understand, the following:

Condition: Inborn errors of amino acid metabolism are a group of diseases in which the metabolism of protein building blocks, known as amino acids, is impaired. Amino acids are found all over the body and are vital to its every day functions. These diseases are usually caused by a genetic defect in an enzyme required for synthesis or breakdown of these amino acids. As a result of this defect, substances may accumulate resulting in interference of normal body functions. These substances can become toxic.

Methodology: This test uses a method called High Performance Liquid Chromatography, or HPLC to measure the amount and type of amino acids in the blood. Based on these results, the particular enzyme defect can be suggested providing a diagnosis of the inborn error of amino acid metabolism.

If your doctor has a clinical suspicion of an inborn error of amino acid metabolism, he/she may request amino acid profile testing to help diagnose the disease.

Meaning of a Positive Test: A positive test result indicates an excess or deficiency of an amino acid(s). This may indicate the presence of an inborn error of amino acid metabolism. Additional testing may be required to confirm this diagnosis or determine prognosis. Diagnosis of an inborn error of metabolism may result in treatment and dietary modifications aimed at improving outcomes.

Meaning of a Negative Test: A negative result does not completely rule out an inborn error of amino acid metabolism in the setting of high clinical suspicion. However, the likelihood of a disease being present in the setting of a negative result is low.

NewYork-Presbyterian
The University Hospital of Columbia and Cornell

***HIGH READING LEVEL**
INFORMED CONSENT FORM FOR AMINO ACID PROFILES

IF NO PLATE, PRINT NAME, SEX AND MEDICAL RECORD NO.

Genetic Counseling: Since a positive test may suggest a genetic defect leading to inborn error of metabolism, you may wish to obtain professional genetic counseling prior to signing this consent form in order to understand the testing and what the results may mean. Also, since a negative test does not completely exclude the possibility of disease, you may still benefit from genetic counseling. You can request genetic counseling through your physician.

Privacy: The results of the tests performed will be included in your medical record and will be available to individuals/ organizations with legal access to your medical record, on a strict "need-to-know" basis, including, but not limited to the physicians and nursing staff directly involved in your care, your current and future insurance carriers, and other people specifically authorized by you to access your medical records. Current New York State law prohibits discrimination by insurance carriers based on the result of genetic tests. The results of your test may be disclosed if such disclosure is ordered by a court.

The Specimen: The blood specimen will be frozen and retained in the lab for up to **60 days**. No other testing will be performed on this specimen. The specimen may only be used for research purposes if specifically authorized to do so by the patient. Otherwise, the specimen will be destroyed after storage with no additional testing. Please initial here if you consent to your sample being used for research purposes after amino acid profile testing _____.

PATIENT CONSENT

Note: If the patient is under eighteen (18) years of age, the permission of the patient's parent or legal guardian must be obtained, unless the patient is married or the parent of a child.

By signing below, I confirm that I fully understand the information provided to me, the benefits and limitations of the test have been explained to me and my questions have been answered.

- ☐ I consent to testing as described above.
- ☐ I decline testing at this time

Name (please print): _____

Signature of Patient/Parent/Legal Guardian/agent: _____

Relationship to Patient: _____

Date: _____/_____/_____ Time: _____ AM/PM

In accordance with New York State Law, I have discussed the testing specified above with the patient/legal guardian. I have discussed the interpretation of the test results and the availability of genetic counseling. I am satisfied that the patient or the patient's legal guardian who signed above understands the information set forth above. This informed consent was signed in my presence.

Name of Person Obtaining Consent (Please print): _____

Signature: _____ MD/NP

Date: _____/_____/_____ Time: _____ AM/PM

Print Name/ID Code: _____