NewYork-Presbyterian

45350

INFORMED CONSENT FOR PLASMA AND URINE METABOLIC PROFILE ANALYSIS

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

Type of Specimen:
Fasting Plasma: Lithium Heparin tube, 1-2ml
Random Urine: Preservative free tube, 5-10 ml.
Sample should be placed in a bag with ice and **immediately** delivered to the laboratory after collection.
Please contact the Biochemical Genetics Laboratory with specific questions: 212-305-6248

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

Please circle the tests that are being requested:

PLASMA AMINO ACID PROFILES **PLASMA ACYLCARNITINE PROFILES**

URINE ORGANIC ACID PROFILES

ICD-10 Code:_____

Requesting Physician:_____

Phone:_____

Pager #:_____

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:
(Name print)

Condition: A variety of chemical compounds (metabolites) exist in the plasma and urine as a result of normal biology, diet, and pharmacological agents (drugs). These chemical compounds are from the normal breakdown (metabolism) of foods that you eat as well as the normal biological processes inside the cell. These processes are mediated by specialized proteins called enzymes. When there are problems with specific enzymes, a specific metabolite or a group of metabolites accumulate in the plasma and urine in a specific pattern. Analysis of plasma and urine may help your doctor determine if a metabolic disease exists and which of the hundreds of possible pathways are affected.

Methodology: This test uses a method which allows for the separation of all the metabolites (chromatography) so that they can be identified and quantified by UV absorbance (HPLC) or Mass Spectrometry (LC-MS/MS or GC-MS) to screen urine and plasma for normal and abnormal metabolites. Based on these results, a particular enzyme defect can be suggested providing a diagnosis of the inborn error of metabolism. If your doctor has a clinical suspicion of an inborn error of metabolism, he/she may request metabolic profile testing to help diagnose the disease.

Meaning of a Positive Test: A positive test result may indicate the presence of a possible inborn error or metabolism. Depending on which metabolites are identified, different diseases are more likely. 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. False positives may occur due to dietary and pharmacological interferences. Your doctor will interpret results of this test against your medical history.

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

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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 urine specimen will be frozen and retained in the lab for **60 days**. Prior to testing for urine organic acid profiles, urine creatinine will be analyzed to determine the correct amount of urine to use for the test. No additional clinical testing will be performed on this specimen. The specimen may only be used for research purposes if specifically authorized to do so by you, 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 metabolite 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