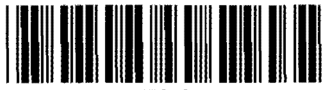
NewYork-Presbyterian
The University Hospital of Columbia and Cornell



*HIGH READING LEVEL

INFORMED CONSENT FORM FOR PLASMA ACYLCARNITINE 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 (or within 2 hours) 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 must also be received with the sample 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: Acylcarnitines accumulate in conditions in which a genetic defect of metabolism exists. Analysis of these chemicals plays an important role in the diagnosis of defects in metabolism of some fats and amino acids.

Methodology: Profiles are obtained using tandem mass spectrometry in Multiple Reaction Monitoring (MRM) mode.

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

Meaning of Test Results:
The results of this testing may
1) indicate that the patient has an inborn error of fatty acid or amino acid metabolism;
2) confirm a clinical diagnosis of the condition;
3) or may have uncertain or minimal clinical significance.

Meaning of a Positive Test: A positive acylcarnitine screen may indicate the presence of an inborn error of fatty acid or amino acid metabolism. Additional testing (enzyme functional assay and/or molecular testing) is generally needed to confirm positive findings of acylcarnitine profiling. My physician has discussed with me how the test will be interpreted, and I understand that I can be referred for genetic counseling, if desired. If I get a positive test result, I may also wish to consider further independent testing or to consult further with my physician.

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Meaning of a Negative Test: A negative result does not exclude the possibility of inborn errors of metabolism in the setting of high clinical suspicion. My physician has discussed with me how the test results will be interpreted and I understand that I can be referred for genetic counseling, if desired.

Genetic Counseling: Since a positive test may suggest a genetic defect leading to inborn error of metabolism, I 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, I may still benefit from genetic counseling. I can request genetic counseling through my physician.

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