



WESTCHESTER
MEDICAL CENTER

BIOCHEMICAL GENETICS LABORATORY
100 Woods Rd.
Valhalla, N.Y. 10595 (914) 493-6541

Consent for Biochemical Genetic Testing.

A specimen is being collected on you/your child for biochemical genetics testing. Testing will involve analysis blood for plasma amino acid analysis and urine for organic acid analysis. These tests look at the body's metabolism and an abnormality in these tests may indicate a possible defect in the metabolism of the body, also known as an inherited metabolic disease (inherited since the majority of them have a genetic cause). The test is being used as a genetic screening test and may lead to the diagnosis of a suspected or as yet unknown genetic disorder.

A more detailed discussion of the testing with a review of expected results can be arranged by genetic consultation. Please ask your physician for a medical genetics consult.

The test results may indicate directly a particular disorder, in which case repeat testing would be indicated for confirmation, though often other tests are indicated for confirmation of the disorder. Due to the nature of the body's metabolism these tests can be normal even in the presence of a particular disorder.

The results of these tests may indicate the presence of many disorders- including disorders in the body's metabolism of carbohydrates, fats and protein,

If a diagnosis is suspected by the test results then certainty depends on the level of the abnormality in the metabolite- such that levels just above normal raise the suspicion of the diagnosis without being diagnostic whereas clearly abnormal levels more strongly suggest the disorder, especially if repeat testing confirms the results.

The test results will only be released to your treating physician or his/ her designee. You may obtain a copy of the results for yourself. Additional disclosure will require signed consent from you.

No additional testing, unless authorized by you will be performed on the specimen. Any remaining sample will be destroyed after 60 days.

Your signature here indicates consent for testing to be sent to the Biochemical Genetics Laboratory.

Patient Name: _____

Signature: _____

Guardian Name: _____

SIGNATURE: _____

Witness Signature: _____

Date: _____