



Seattle Children's
HOSPITAL • RESEARCH • FOUNDATION

Biochemical Genetics Laboratory OC.8.720
4800 Sand Point Way NE, Seattle, WA 98105

Consent for Prenatal Testing by Biochemical Testing

I understand that the purpose of this testing is to determine if my child could be affected with

This testing should be done in conjunction with genetic counseling. I understand that this test does not determine carrier status.

This enzyme testing is specific for the enzyme defect listed above, and does not rule out other conditions, or similar conditions caused by different enzyme defects. Normal enzyme activity does not guarantee my child's general health.

The nature and accuracy of biochemical analysis has been explained to me. I understand that this testing may not provide a 100% definitive diagnosis with regard to disease status. Because of the wide range of physiologic enzyme activity in cultured cells and the occasional presence of pseudo-genes that can cause reduced enzyme activity in normal individuals, there is a small chance of overlapping enzyme activity between unaffected carriers of disease and affected individuals. In other words, there is a small risk that a carrier can have a result that looks affected or an affected individual can have a result that looks like a carrier. With any laboratory testing, there is a small possibility that the test will not work properly, cells will not grow, or an error will occur. There are rare occasions, when the mother's cells may also be present in the sample sent to us. This is called *maternal cell contamination*. If this occurs, the test may yield a false result and a new prenatal sample will be required. Therefore, we require a blood sample from the mother so we can see if there are maternal cells in the test sample.

The laboratory will make every effort to report results within the estimated turn around time, but cannot accept responsibility for delays.

Due to the complexity of this testing and the important implications of the test results, results will be reported only through the provider or genetic counselor designated below.

I have carefully read and understand the above, have had my questions answered, and hereby consent to provide cultured chorionic villus cells or amniotic fluid cells, as appropriate, and a maternal blood sample for testing.

Name of patient (please print)

Signature of patient

Date

Signature of provider/genetic counselor

Date