Clinic *for* Special Children

Informed Consent for Molecular Genetic Testing

Patient					CSC ID number	
Street address						
City		State		Zip code		
Telephone		Birthdate				
Father		Mother				
Paternal grandmother (with maiden name)		Maternal grandmother (with maiden name)				
Paternal grandfather		Maternal grandfather				
Spouse (with maiden name)		Spouse's birthdate				

The diagnosis of a genetic disease or the identification of people at risk for having a child with a genetic disease requires the testing of DNA for the presence of an abnormal gene. You and/or your healthcare provider are requesting the Clinic for Special Children (CSC) to perform a genetic test on you or your child. The purpose and accuracy of this test has been explained to you and your questions have been satisfactorily answered.

Development and performance of this testing was determined by the CSC. This clinical testing has not been approved by the FDA; however, the FDA has determined that such approval is not necessary. Some reagents used in this testing are produced for research purposes only. There is always a chance that an error may occur (including, but not limited to, sample contamination and sample misidentification). A negative test result does not necessarily exclude a genetic disease. Results of genetic testing should be considered with the results of other laboratory testing as well as clinical evaluation. The results of these tests will be handled in the standard medically confidential manner.

You and/or your healthcare provider acknowledge permission to (1) obtain about 3 ml of blood from a vein (2) isolate DNA from this sample, (3) perform the requested diagnostic tests (if any), and (4) store the DNA sample at the Clinic for Special Children. By signing this form, you acknowledge that any remaining DNA may be used further for quality-control purposes or additional research. Your name or other personal identifying information will not be used in or linked to the results of any studies or medical publications. However, you have the right to learn of any medically significant findings identified in the course of that process. Please select your preference below:

☐ **I DO** *or* ☐ **I DO NOT** agree to be re-contacted for future research studies relevant to the CSC's mission. I understand that my decision to opt-out of such follow-up contacts will not affect my ability to obtain testing or receive medical care at CSC.

☐ **I DO** *or* ☐ **I DO NOT** want to be informed of incidental fir.dings (medically actionable findings not related to the indication for testing)

Plain Insight Panel (PIP) ONLY: By choosing to do the Plain Insight Panel, you are electing to learn the following genetic information from the test: 1. Genetic variants associated with *known conditions* manifesting in childhood or adulthood; 2. Genetic variants *predicted to cause conditions* that have not yet been seen in Plain patients at the Clinic; 3. Genetic variants that may impact health by increasing the severity of a condition *only* when in combination with another genetic or non-genetic factor, but are likely insufficient to cause the disease alone.

Signature of patient *or* parent/legal guardian **Date**

Alternate consent: I, the health care provider requesting the above testing, have explained the benefits and limitations of genetic testing to the patient and/or their legal guardian and have received verbal consent to order genetic testing.

Signature of healthcare provider **Date**

Laboratory Requisition

Clinic *for* Special Children
535 Bunker Hill Road, PO Box. 128. Strasburg, PA 17579 **T** 717.687.9407 **F** 717.687.9237

1 Patient

_____ _____
Name # Date of Birth

_____ _____
Address # Phone number

If pregnant, what is the expected due date? _____

2 Sample (Ship Mon.-Thurs. ONLY by overnight delivery. Call for emergent samples at other times)

☐ Peripheral blood ☐ Cord blood ☐ Filter card

3 Testing Requested

☐ Single targeted mutation tests *(Please see test list at http://www.clinicforspecialchildren.org)*

_____ _____ _____
Disease (e.g., MSUD) *Gene (e.g., BCKDHA)* *Variant (e.g., c.1312T>A)*

_____ _____ _____
Disease *Gene* *Variant*

Indication for targeted testing

- ☐ Carrier testing
  - ☐ *Clinically normal individual with no family history*
  - ☐ *Family history of the condition*
  - ☐ *Spouse with family history of condition*
  - ☐ *Spouse is a known carrier for condition*
- ☐ Diagnostic testing
  - *Clinical indication* _____

☐ Chromosomal microarray *(Peripheral or cord blood only)*

Clinical indication _____

☐ Plain Insight Panel™ *(Peripheral or cord blood only)*

- ☐ Carrier screening - healthy adult with no specific personal medical concerns
- ☐ Diagnostic testing - identify genetic basis for this patient's medical issues
- ☐ Parental screening - parental testing to identify the genetic basis for their child's health concerns

4 Reporting

_____ _____
Requesting physician/ midwife/ counselor *Institution*

_____
Address

_____ _____ _____
Phone number ☐ # Fax number * ☐ Email *

Billing

_____ _____
Name *Institution*

_____
Address

_____ _____ _____
Phone number # Fax number *Email*

 # These items are REQUIRED. Failure to provide proper documentation may result in delayed testing or rejection of sample.*POSITIVE diagnostic results will be reported by phone to requesting provider. Please indicate preference for reporting NEGATIVE results by checking the appropriate box (fax or email).