UPSTATE
UNIVERSITY HOSPITAL

CONSENT FOR CHROMOSOME SNP MICROARRAY TESTING

Patient Name: _____ MR#: _____

Account #: _____ DOB: _____ Date: _____

Your health care provider has recommended that you consider having a genetic test called Chromosome SNP Microarray (CSM) which may confirm a disease diagnosis or indicate that you (or your child) is a carrier or has an increased risk of expressing a genetic disease. **You may also wish to obtain genetic counseling prior to signing this form.**

1. **What is Chromosome SNP Microarray Testing?** Approximately 10cc of blood will be drawn by your physician or a professional phlebotomist. The DNA (genetic material of the cell) will be isolated and evaluated. The Chromosome SNP Microarray (CSM) testing analyzes the whole genome for gains and losses (copy number change) of genetic material (DNA). This test reveals the same type of information as a standard karyotype analysis, but the resolution is much higher and the information obtained is much more detailed. The Cytogenetics Laboratory uses a microarray developed by Affymetrix, Inc. which is called the Cytoscan HD.
2. **What is the purpose of CSM?** This test provides a high resolution scan of an individual's genetic material to determine if there are gains or losses of genetic information that may explain abnormal clinical findings identified by your physician. The results may confirm a disease diagnosis or indicate that an individual is a carrier or has an increased risk of expressing a genetic disease. Identification of an abnormality may be helpful in managing your health care and/or determining the best treatment for your condition. In some cases, no clinically significant anomalies will be found, thus the test will provide no direct information about the cause of your medical condition.
3. **What are the limitations?** Microarray analysis was developed to detect copy number gain or loss in genomic DNA. CSM testing can only identify changes (aneuploidies, duplications, deletions, mosaicism) for the specific regions included in the microarray. It cannot detect balanced rearrangements (translocations, inversions, insertions), point mutations, or imbalances of regions not surveyed by the microarray. CSM may not detect very small gains, losses, or low levels of mosaicism that are below the resolution of the array that is used. While results of this test are over 99% accurate, errors such as false positives or false negatives may occur. Furthermore, a negative result (failure to detect an abnormality) does not exclude the possibility of a pathogenic mutation in the regions interrogated by this assay.
4. Interpretation of test results is based on currently known genetic abnormalities (mutations) that have been directly associated with disease. This test may identify changes in your genome that have not been correlated with disease, so these findings would be reported as having no known consequences. It is possible that in the future one or more of these genetic anomalies may become diagnostically important, but it is not possible to predict that at this time.
5. Correct interpretation is also dependent on the accuracy of the clinical and family history information submitted with the sample. However, this type of genetic testing can sometimes reveal that the true biological relationship between family members is not consistent with the history provided. For example, non-paternity may be detected, which means that the stated father of an individual is not the true biological father.
6. **Additional testing may be necessary** for confirmation of positive results. By signing this consent, you agree to allow the laboratory to perform supplementary testing, such as fluorescence in situ hybridization (FISH), to confirm the CSM results. In some cases, a full interpretation of the data may require CSM studies on other family members (usually both parents) to determine if specific gains or losses of genetic material are inherited. These data may help to determine if the changes are benign or if other family members may be at risk of acquiring the same disease. If additional testing is appropriate, you will be contacted by your referring physician.
7. **What will happen to the cells once the test is complete?** No clinical tests other than those authorized will be performed. The sample will be discarded within 60 days after testing is complete, unless an additional consent for longer specimen retention is obtained.
8. I agree that my (or my child's) sample may be used for test validation or education after personal identifiers have been removed. In this case, the sample may be stored indefinitely. If I do NOT wish to allow use of the sample for test validation or education, I will check this box ☐.

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9. **What will happen to the test results?** All results and records shall be treated as confidential. The results of the genetic testing will be sent to the health care professionals who requested the testing.
10. **How will I obtain results from the test?** Please contact your physician or designated genetic counselor to obtain test results and interpretation of the findings.
11. I specifically authorize my test results to be provided to my health care insurer or health maintenance organization to the extent that they are reasonably required for the purposes of claims administration.
12. The New York State Genetics Testing Confidentiality Law Article 7, Section 79-1 requires that all individuals be informed of the nature of the genetic testing being requested. I confirm that my physician has explained to me the reason for this testing, the procedure, the possible results and implications thereof, and the approximate time for results to become available. By my signature on this informed consent, I confirm that I have had an opportunity to obtain professional genetic counseling prior to signing this informed consent. My questions have been answered and my concerns addressed to my satisfaction. I understand that I may ask for further information or additional counseling and it will be provided to me.
13. I have read this entire document and understand its contents.

If you have any questions about the test which will be performed, you may contact the Cytogenetics Laboratory at 315-464-4716.

Please indicate the name and address of any other physicians to whom you wish a copy of the report to be sent.

Name: _____

Address: _____

_____

My signature below indicates that the above information has been explained to me and that I give consent for chromosome SNP microarray testing.

Date: _____

Signature of Patient or Parent/Guardian, if patient is a minor: _____

Patient's name (printed): _____

Signature of Referring Physician/Genetic Counselor: _____

Referring Physician/Genetic Counselor Name (printed): _____

UPSTATE
MEDICAL UNIVERSITY
Department of Pathology
Cytogenetics Laboratory

DOCUMENTATION OF MEDICAL NECESSITY FOR MICROARRAY TESTING
TO BE COMPLETED BY ORDERING PROVIDER

Patient Name: _____ Date of Birth: _____

Insurance Provider: _____ Policy ID #: _____

This patient is currently being cared for by the following physician, who has ordered **Microarray testing**, CPT Code(s) 81229:

Referring Physician: _____

Phone: _____ Fax: _____

Indication(s) for Testing: _____

_____

Previous Genetic Testing: _____

Medical Necessity Notes (attach additional pages if needed): _____

_____

_____

☐ HOLD CELLS, DO IF KARYOTYPE IS NORMAL.

ICD9 Codes (check all that apply):

☐ Delayed Developmental Milestones (783.42)
☐ Dysmorphic Features (796.4)
☐ Developmental Delay (315.9)
☐ Seizure Disorder (345.9)
☐ Multiple Congenital Anomalies (759.7)
☐ Autism Spectrum Disorder (299.0)
☐ Failure to Thrive Child (Child 783.41; Newborn 779.34) (circle one)
☐ Clarify Abnormal Chromosomes (758.89) Provide results and a copy of karyotype.
☐ Other: _____

_____

_____

Making a diagnosis is essential for this patient's current healthcare and ongoing medical management.

Signed: _____ Date: _____

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PRE-AUTHORIZATION OFFICE USE ONLY

PLEASE FORWARD PRE-AUTHORIZATION TO:

☒ Cytogenetics Laboratory/Upstate Medical University
750 East Adams Street, Syracuse, NY 13210
Phone: (315) 464-4716 Fax: (315) 464-4718, Attention: Lori/Cindy
Email: plaistel@upstate.edu and/or lafountc@upstate.edu

Pre Pre-authorization #: _____