www.CytoGenX.com

1212 Rte 25A
STONY BROOK, NY 11790
1-888-GENE-MED

INFORMED CONSENT AND RELEASE FOR MOLECULAR CYTOGENETIC STUDIES: MICROARRAY

I voluntarily consent for microarray analysis to be performed on my specimen or my child's specimen.

Description and purpose of Array CGH + SNP: The chromosome constitution of a patient or fetus with a suspected DNA copy number change is compared to the DNA of a known reference control set of normal individuals. The purpose of this test is to determine whether my (or my child's or my fetus') sample has changes in the DNA copy number that may explain the clinical presentation. This test will reveal major chromosome abnormalities and sub-microscopic chromosomal imbalances and is considered to be greater than 99% accurate.

Chromosome abnormalities and sub-microscopic imbalances may be associated with:
Developmental delay
Congenital abnormalities
Infertility
A history of miscarriage
Embryonic and fetal death
Short stature
A family history of a chromosome abnormality, mental retardation or birth defects.

In the event a chromosomal aberration is identified, genetic counseling is highly recommended to explain the meaning of the result as well as discuss options for clinical management.

The following points have been explained to my satisfaction by a qualified health professional and my signature below indicates that I understand the benefits, risks, and limitations of this testing and accept them:

- I have been offered the option of receiving genetic counseling before and after the test.
- The nature and scope of the conditions tested for have been explained to me and I have been given access to a list of these conditions.
- If the test is "positive," this would be an indication that I (or my child's or my fetus) may be predisposed to or have the specific condition(s) tested for, and I may wish to consider further independent testing, consult my physician, or pursue genetic counseling.
- A normal ("negative") test result does not exclude the possibility that I/my child/my fetus may have a genetic condition which is not surveyed by the microarray analysis. Microarray analysis assesses DNA copy number changes and does not rule out genetic disorders caused by single gene mutations.
- I understand that when the microarray testing does not show a genetic change (a "negative test result"), the chance that I am a carrier of or affected with one of the conditions tested for is reduced, but not eliminated. I understand that the genetic conditions included in this testing panel may be caused by more than one genetic mechanism; therefore there is still a chance to be affected because current testing cannot find all the possible changes within the genome.
- The test may be unsuccessful on rare occasions.
- If a gain or loss is reported, the change will be confirmed by additional testing, such as standard chromosome analysis or Fluorescence in situ Hybridization (FISH). Additional tests may be necessary to fully interpret an abnormal result.
- Genetic testing of parents can reveal non- paternity. Misinformation regarding the true biological relationships of the parents can result in significant reporting errors.
- This test may identify consanguinity (a close blood relationship or incest) or non-paternity.
- Accurate interpretation of test results is dependent upon the accuracy of the information submitted with the sample. This includes the fetus' clinical findings, if any, and the family medical history.

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- It is possible for the array to find a gain or loss of chromosome material that is of uncertain significance. In the majority of such cases the gain or loss is inherited which is less worrisome, and in about 1% of cases the gain or loss is found only in the fetus. Genetic counseling is highly recommended in such situations.
- In rare circumstances, findings may be suggestive of a condition different than the diagnosis that was originally considered.
- Chromosomal microarray cannot detect balanced chromosomal rearrangements, point mutations or imbalances of regions not included on the microarray.
- The rate of birth defects and mental retardation in the general population ranges from 3-5%. Therefore, a normal chromosomal microarray result does not guarantee a healthy child.
- Chromosomal microarray may identify a chromosomal abnormality. Identifying this abnormality may be useful in directing additional care, evaluation or monitoring for your pregnancy or for your child after delivery.
- The specimen may be forwarded, by the CytoGenX Laboratories, to another accredited laboratory for testing if the CytoGenX Laboratories cannot perform the requested test.

I have been given the opportunity to ask questions about the ordered tests and told how I will receive the test results. I understand that my results will be disclosed to the ordering physician and/or genetic counselor. I have received genetic counseling prior to signing this form and have been told that counseling is available after the test is performed. It is the responsibility of the referring clinician to understand the utility and limitations of the testing ordered, and to educate the patient regarding these issues in relation to their care. Due to the complexity of genetic testing and potential implications of test results, results will be reported only to the ordering provider.

No tests other than those authorized shall be performed on the biological sample and the sample will be destroyed at the end of the testing process or not more than sixty days after the sample was taken, unless a longer period of retention is expressly authorized in the consent or unless consent is given for additional purposes.

Upon completion of testing, a portion of my specimen (or my child's or my fetus') may be made anonymous and used for test validation, research or educational purposes. Once the material has been made anonymous, its original source can no longer be identified.

I do not wish to allow my specimen (or my child's or my fetus' sample) to be de-identified and used for test validation, research or education. Therefore, I am checking this box to indicate that the sample should be used only for the test specified above and will be destroyed after sixty days. ☐

Print name of patient/authorized representative	Date	Print name of person obtaining consent	Date
Signature of patient/authorized representative	Date	Signature of person obtaining cons	Date