



45350

**INFORMED CONSENT AND RELEASE FOR CONVENTIONAL
AND MOLECULAR CYTOGENETIC STUDIES, AND
CYTOGENOMIC MICROARRAY STUDIES FOR
CONSTITUTIONAL GENOMIC DNA COPY NUMBER
VARIATION ANALYSIS**

IF NO PLATE, PRINT NAME, SEX AND MEDICAL RECORD NO.

Informed consent and release for conventional and molecular cytogenetic studies, and microarray studies for constitutional genomic DNA copy number variation analysis

I voluntarily consent for the following tests to be performed on my /my child's/my fetus's _____ specimen.

- ☐ Conventional cytogenetic analysis ☐ Molecular cytogenetic analysis (FISH)
- ☐ Cyto genomic Microarray testing

Description and purpose of the tests:

Chromosome and sub-microscopic chromosome abnormalities may be associated with developmental delay (mental retardation, behavioral and /or learning disabilities), congenital abnormalities, infertility, history of miscarriage, embryonic and fetal death, short stature, and family history of a chromosome abnormality.

Cytogenetics and fluorescence in situ hybridization (FISH): The chromosome constitution of my specimen will be analyzed by growing the cells in culture and examining the chromosomes under the microscope. This test will reveal major chromosomal abnormalities and is considered to be greater than 99% accurate. In some cases, a preliminary FISH analysis may be performed on uncultured cells and this test will reveal information on a limited subset of chromosome abnormalities.

Cytogenomic Microarray (CMA) testing: is a molecular cytogenetic test that is performed on genomic DNA extracted from peripheral blood and compares the DNA of a patient to the DNA of a known reference control set of normal individuals. Microarray test is capable of detecting allelic imbalances (gains and losses) and copy number neutral abnormalities such as loss of heterozygosity (LOH) and region of homozygosity (ROH or AOH) that can be associated with uniparental disomy (UPD) or consanguinity, both of which may pose increased risk for autosomal recessive conditions. Microarray has higher resolution than conventional cytogenetics and has more comprehensive coverage than FISH. The purpose of this test is to determine whether my sample / my child's or my fetus sample has changes in the DNA copy number that may explain the clinical presentation.

The Cytogenetics Laboratory at NYPH/WCM uses single nucleotide polymorphism (SNP) array that spans all chromosomes and includes all known microdeletion and microduplication syndromes. Conventional cytogenetic analysis or microarray on parental DNA will be necessary if microarray detects a gain or loss of genetic material and is of unknown clinical significance. Even though most of the inherited changes are not likely to be pathogenic, in some cases one of the parents may be carriers of a gain or loss of the genome or carriers of a balanced chromosome rearrangement which may have been transmitted to the child in an unbalanced form.

Microarray testing will not detect low level mosaicism (<20%), balanced rearrangements, specific gene point mutations or rearrangements below the resolution of the array.

In the event of a chromosomal aberration is identified by conventional cytogenetics, FISH or microarray, genetic counseling is recommended to explain the meaning of the results as well as to discuss options for clinical management.

The following points have been explained and I understand and accept them:

1. I have the option of receiving genetic counseling before and after the procedure.
2. Tissue culture from the cells of any particular sample may be unsuccessful or the chromosomes preparations may be of poor quality and unusable (Conventional Cytogenetics and FISH). Cytogenomic microarray testing may be unsuccessful on rare occasions.
3. Conventional Cytogenetics, FISH assays, and Cytogenomic microarray will not detect very small deletions, duplications and subtle rearrangements or low level mosaicism. In rare cases, the test may provide results that are difficult to interpret in terms of clinical outcome. I understand that while the results from these tests are highly accurate, infrequent errors may occur.
4. Additional testing may be needed to confirm or refine the interpretation of test results.

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5. I have the option if my or my child's or my fetus's test is "positive" I may consider further independent testing and pursue genetic counseling.
6. A normal test result does not exclude the possibility that I/my child/my fetus may have a genetic condition which is not evaluable by conventional Cytogenetics, FISH, and Cytogenomic microarray testing.
7. I understand that all three tests do not rule out genetic disorders caused by single gene mutations.
8. The specimen may be forwarded, by the NewYork Presbyterian Hospital laboratories, to another accredited laboratory for testing if the NewYork-Presbyterian Hospital laboratories cannot perform the requested test.
9. The test results will be a part of my medical record and will be available to physicians and genetic counselors involved in my care.
10. I have been given the opportunity to ask questions about the ordered tests and told how I will the get the test results.
11. I would like to receive the results of **all clinically relevant findings** from the Cytogenomic microarray test, even if they are unrelated to the reason that the study was initially ordered. The ordering doctor/health care provider or his/her designee can report the results to me. ☐ **Yes** ☐ **No** Initials: _____
12. No test other than those authorized shall be performed on the biological sample and the sample will be discarded 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 testing purposes.
13. Upon completion of testing a portion of my specimen may be made anonymous and used for test validation or research purposes. Once the material has been made anonymous, its original source can no longer be identified. I give consent for item 13. ☐ **Yes** ☐ **No**

Patient Signature/Authorized Representative Print Name Date ____/____/____ Time ____^{AM}____^{PM}

Person Obtaining Consent Print Name Date ____/____/____ Time ____^{AM}____^{PM}