Northwell Health® Laboratories

PRENATAL CYTOGENETICS TEST REQUISITION

450 LAKEVILLE RD., SUITE M50
LAKE SUCCESS, NY 11042

OUTLINED AREAS MUST BE COMPLETED

AFFIX TO SPECIMEN CONTAINER

PATIENT

PATIENT IDENTIFIER			
NAME, LAST (Please Print)	FIRST	M.I.	
BIRTHDATE	M/F	DATE/TIME COLLECTED	
STREET	PHONE #		
CITY	STATE	ZIP	

BILLING

INSURANCE CARRIER NAME	ADDRESS	
INSURED NAME	INSURED ID#	PT. RELATIONSHIP TO INSURED: ☐ Self ☐ Spouse ☐ Dependent
GROUP # or NAME		☐ INSURANCE CHANGE
☐ MEDICARE #	☐ MEDICAID #	☐ SELF-PAY

RACE/ETHNICITY ☐ NATIVE AMERICAN ☐ ASIAN ☐ AFRICAN-AMERICAN ☐ CAUCASIAN
☐ ASHKENAZI JEWISH ☐ PACIFIC ISLANDER ☐ HISPANIC ☐ OTHER _____

I attest that this patient has been informed about and has given consent for the test(s) I have ordered below under applicable law.
PHYSICIAN SIGNATURE: (required)

X

REPORT COPY TO:

DIAGNOSIS		DX CODE
DX CODE	DX CODE	DX CODE

ORDER COMMENTS

INFORMED CONSENT	Patient has read the appropriate consent form on the following pages and has been informed of the required information regarding testing for inherited genetic disorders or predisposition. Patient Signature: _____ Healthcare Provider Signature: _____ Date: _____

For questions please contact NSUH Laboratory phone: 516-562-3898; fax: 516-562-2691
Hours of Operation: Monday-Friday, 8am-5pm

IMPORTANT: Please have the patient sign the **Informed Consent for Cytogenetic/Molecular Cytogenetic Testing Form** (page 2). Informed consent is required for all genetic samples.

SPECIMEN TYPE ☐ **Amniotic fluid** ☐ **CVS** ☐ **Percutaneous umbilical cord blood (PUBS)**
Date & Time Specimen Collected: ___/___/____ ____ : ____ **GA on date of procedure:** ____ wks ____ days by sono

CLINICAL INFORMATION & INDICATION **Type of Pregnancy:** ☐ Singleton ☐ Twins ☐ Triplets ☐ Other _____

☐ Advanced maternal age (> 35)
☐ Abnormal Maternal Serum Screen: _____
☐ Abnormal NIPS: _____
☐ Abnormal ultrasound _____
☐ Parental chromosome abnormality _____
☐ Other _____

TEST(S) REQUESTED (performed at NSUH Cytogenetics Lab)
☐ **Chromosomes** ☐ **Prenatal Aneuploidy FISH (13, 18, 21, X and Y)**
☐ **Grow cells for:** _____
☐ **Hold cells for:** _____

SEND OUT TESTS **PLEASE FAX ADDITIONAL REPORT TO GENETIC COUNSELOR:(____)_____**

☐ **AFP NTD** Amniotic Fluid AFP
☐ **ACHE** **AChE Only**
☐ PREARRAY **Prenatal Targeted Chromosome Microarray***
☐ PrenatSNP **High Resolution Chromosome MIcroarray***
☐ **Maternal Cell Contamination Studies** (10 ml EDTA) send to: _____
☐ **Noonan Syndrome Gene Sequencing Panel** send to: _____
☐ **Smith-Lemli-Opitz Syndrome 7-DHC** (Kennedy Krieger)
☐ **Amniotic Fluid** ☐ **CMVPCRAMN** ☐ **ToxoPCR** ☐ **MISC ParvoPCR**

***Please include blood specimen from parents of the proband if available**
(1 EDTA lavender top tube) ☐ Inciuded ____ mother ____ father
☐ Other: _____

ADDITIONAL INSTRUCTIONS: _____

For Lab Use Only-Specimen Processing Data
Date Received: _____ / ____ /_____ Time Received: _____ : _____
Specimen Quantity: _____ Specimen Quality: _____

PATIENT INFORMATION SHEET & INFORMED PATIENT CONSENT

CYTOGENETIC & MOLECULAR CYTOGENETIC TESTING

What is cytogenetic, fluorescence *in situ* hybridization (FISH), and chromosome microarray testing?
Chromosome disorders form a major category of genetic disease and account for a large proportion of congenital malformations, intellectual disability, and fetal loss. **Conventional cytogenetic testing or routine chromosome analysis (karyotyping)** is the analysis of human chromosomes, their structure and their inheritance. This testing is utilized to detect numerical and/or structural chromosome abnormalities. **FISH** is a rapid and sensitive technique that complements routine chromosome analysis. It uses specific fluorescent-tagged DNA probes to detect and localize the presence or absence of specific DNA sequences. **Chromosome microarray** is a molecular cytogenetic test that has the ability to detect smaller deletions/duplications in addition to the larger chromosome imbalances routine chromosome analysis can detect.

What are the limitations of the test(s)?
Chromosome analysis does not routinely detect subtle structural changes or microdeletion/microduplication syndromes. **Chromosome microarray** does not detected balanced chromosome rearrangements, and may identify variants of uncertain clinical significance (VUS), in which cases parental follow up may be indicated. Neither test can detect low levels of mosaicism, or conditions with Mendelian, multifactorial or environmental causes. **FISH** is a targeted approach; the information generated is specific to the probes used and may give normal test results in some patients with other genetic causes. FISH is considered an adjunct to routine chromosome analysis performed concurrently. ***No irreversible decisions about a pregnancy should be made on the basis of FISH results alone.*** As with any laboratory test, there is a small possibility of failure or error.

What Is required to perform this test?
All three tests can be performed on several different specimen types based on the indication, including prenatal specimens (amniotic fluid, chorionic villi, and umbilical cord blood), and postnatal specimens (products of conception (POC), skin biopsy, and peripheral blood). An accurate clinical history is critical for proper interpretation of the results.

When should I expect test results?
Chromosome analysis: prenatal specimens approximately 7-14 days/ peripheral bloods ~ 2 weeks/ POCs ~2-3 weeks
FISH: prenatal specimens approximately 24-48 hours/ peripheral bloods 7-10 days
Chromosome microarray: approximately 2 weeks

Who will contact me regarding test results?
Test results will be forwarded to your physician and genetic counselor. A positive result is an indication that you may be predisposed to or have the specific disease or condition tested for and may want to consider further independent testing, consult your physician or pursue genetic counseling. A recommendation for additional testing on the patient and/or the parents/other family member will be made in the event of an abnormal chromosomal finding or VUS to determine whether a specific finding was inherited.

Confidentiality of test results
The test results will be disclosed to the requesting physician(s) and to associated medical personnel only. To the extent permitted by law, all of the records, findings and results of this test are confidential and will not be disclosed to another physician without the written authorization of the patient/guardian.

Specimen retention
The specimen will be discarded within 60 days of collection, at the end of testing and after final reporting, unless additional testing is requested. No tests other than those authorized will be performed on the sample. Any residual specimen not used for diagnostic testing may be retained for use by the laboratory for the purposes of quality control, training purposes. or for research with patient signed consent (see Residual Material section).

INFORMED PATIENT CONSENT
As required by Section 79-1 of the Civil Rights Law, written informed consent of the individual being tested should be obtained by the laboratory prior to testing for constitutional genetic analysis by chromosome or by DNA study. The individual may wish to obtain professional genetic counseling prior to signing the informed consent.

I have received/read the information regarding **Cytogenetic and Molecular Cytogenetic testing** and hereby give my consent to perform the test(s). I understand that a positive result may not result in a genetic condition, but may predispose to it. Such a result may require genetic counseling, further testing and/or further physician consultation. A negative result does not rule-out a genetic condition. The test may give a false negative result due to changes not detectable by the method and/or reagents used. The results of the test(s) are confidential and will be disclosed to requesting physicians, their staff and those legally authorized. I give my consent to the above testing.

_____	_____	_____	_____	
Patient/Agent/Relative/Guardian* (Signature)	Date	Time	Print Name	Relationship if other than patient

_____	_____	_____
Telephonic Interpreter's ID #	Date	Time

OR

_____	_____	_____	_____
Signature: Interpreter	Date	Time	Print: Interpreter's Name and Relationship to Patient

_____	_____	_____	_____
Witness to signature (Signature)	Date	Time	Print Witness Name

RESIDUAL MATERIAL
I consent to having my specimen retained for greater than 60 days for future testing or the use by the laboratory for the purposes of quality control and/or training purposes or for research related to, but not limited, to genetic disease pursuant to a research protocol approved by an institutional review board. I understand this is not a DNA banking facility and there are no guarantees a specimen will be remaining for future testing. If used for quality control and/or training purposes or research, all identifying information will be permanently stripped from the sample. I hereby give my consent to the above.

_____	_____	_____	_____	
Patient/Agent/Relative/Guardian* (Signature)	Date	Time	Print Name	Relationship if other than patient

* The signature of the patient must be obtained unless the patient is an unemancipated minor under the age of 18 or is otherwise incapable of signing.
** The witness to signature may be the physician or genetic counselor.