Northwell Health® Laboratories

Core Account #: _____ **Ordering MD**

□_____□_____ □ _____

PRENATAL CYTOGENETICS TEST REQUISITION

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.

PATIENT INFORMATION

Patient Name: _____ Date of Birth: ____/____/_____ Age _____ Sex: ☐ Male ☐ Female

Address: _____ Medical Record Number: _____

City, State, ZIP _____ Telephone: (_____) _____- _____

PHYSICIAN INFORMATION

Referring Physician: _____ Physician Phone: (_____) _____- _____ Fax: (_____) _____ - _____

Genetic Counselor: _____Counselor Phone: (___)_____Fax: (___)_____

PHYSICIAN SIGNATURE OF CONSENT

I attest that the patient specified above and/or their legal guardian has been informed of the benefits, risks, and limitations of the laboratory test(s) requested. I have answered this person's questions and have obtained informed consent from the patient or their legal guardian for this testing.

Physician Signature _____ **Print Name** _____ **Date / Time** ___/___/___ ___:___

BILLING INFORMATION

Insurance Carrier: _____ ID # _____ ICD-10 Code: _____, _____, _____

Insured Name _____ Group #_____ Relationship to Insured: ☐ Self ☐ Spouse ☐ Dependent

ASSIGNMENT AND RELEASE: I hereby authorize my insurance benefits be paid directly to the provider and I understand that I am financially responsible for uncovered services. I also authorize the release of any information required to process the claim.

Signature _____ **Print Name** _____ **Date / Time** ___/___/___ ___:___

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)

☐ **Grow cells for:** _____

☐ **Chromosomes** ☐ **Prenatal Aneuploidy FISH (13, 18, 21, X and Y)** ☐ **Hold cells for:** _____

SEND OUT TESTS

PLEASE FAX ADDITIONAL REPORT TO GENETIC COUNSELOR fax: _____

☐ **AFP NTD (Labcorp)**

☐ **AChE (Labcorp)**

☐ **Prenatal Targeted Chromosome Microarray** (GeneDx 410)

☐ **High Resolution Chromosome Microarray** (GeneDx 460)

Please include blood specimen from parents of the proband if available (1 EDTA lavender top tube) ☐ Included _____ mother _____ father

☐ **Maternal Cell Contamination Studies** (10 ml EDTA) send to: _____

☐ **Noonan Syndrome Gene Sequencing Panel** (GeneDx 357)

☐ **Smith-Lemli-Opitz Syndrome 7-DHC** (Kennedy Krieger)

☐ **Amniotic Fluid** □ **CMV** □ **Toxo** □ **Parvo PCR** (Eurofins Viracor)

☐ Other: _____

ADDITIONAL INSTRUCTIONS _____

_____

For Lab Use Only-Specimen Processing Data

Date Received: _____/_____/_____ Time Received: _____:_____

Specimen Quantity: _____ Specimen Quality: _____

Northwell Health® Laboratories

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 # **OR**	Date / Time		
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.