

OUTLINED AREAS MUST BE COMPLETED

P A T I E N T	PATIENT IDENTIFIER			
	NAME, LAST (Please Print)		FIRST	M.I.
	BIRTHDATE	M/F	DATE/TIME COLLECTED	
	STREET		PHONE #	
	CITY	STATE	ZIP	
B I L L I N G	INSURANCE CARRIER NAME		ADDRESS	
	INSURED NAME		INSURED ID#	PT. RELATIONSHIP TO INSURED: ☐ Self ☐ Spouse ☐ Dependent
	GROUP # or NAME		☐ INSURANCE CHANGE	
	☐ MEDICARE #		☐ MEDICAID #	☐ SELF-PAY

AFFIX TO SPECIMEN CONTAINER
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

ORDER COMMENTS

Genetics Counselor name: _____
Fax additional reports to Genetic Counselor (_____) _____

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 ____ / ____ / ____ : ____

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

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

CLINICAL INFORMATION & INDICATION	Type of Pregnancy: ☐ Singleton ☐ Twins ☐ Triplets ☐ Other _____
☐ Advanced maternal age (≥35)	☐ Abnormal ultrasound _____
☐ Abnormal Maternal Serum Screen: _____	☐ Parental chromosome abnormality _____
☐ Abnormal NIPS: _____	☐ Other _____

TEST(S) REQUESTED	☐ Grow cells for: _____
☐ Chromosomes ☐ Prenatal Aneuploidy FISH (13, 18, 21, X and Y)	☐ Hold cells for: _____
☐ AFP AF AFP Amniotic Fluid	

SEND OUT TESTS	☐ CMVPCRAN CMV PCR, Amniotic FL
☐ PrenatSNP High Resolution Chromosome Microarray*	☐ PARVOPCRAN Parvovirus PCR, Amniotic FL
☐ Maternal Cell Contamination Studies (10 ml EDTA) send to: _____	☐ TOXOPCRAN Toxoplasma PCR, Amniotic FL
☐ ACHF AF AChE, Amniotic Fluid With Reflex to Fetal Hemoglobin	☐ PN SkeIDys Prenatal Skeletal Dysplasia
☐ PreNoonan Prenatal Noonan Spectrum Panel	☐ Other: _____
☐ Prenatal Targeted Variant Testing - send to: _____ Gene: _____	
Variants: Maternal _____ Paternal _____	
Please include parental bloods if available (1 EDTA lavender top). Included ☐ Maternal ☐ Paternal: Name: _____ DOB: _____	

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 detect 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
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* 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.