



Core Account #: _____ Ordering MD

☐ _____ ☐ _____ ☐ _____**POSTNATAL CYTOGENETICS TEST REQUISITION****Peripheral Bloods:** for questions please contact LIJ Laboratory phone: 718-470-7065; fax: 718-470-1034**POC/Skin Biopsies:** for questions please contact NSUH Laboratory phone: 516-562-3898; fax: 516-562-2691

Hours of Operation for both laboratories: 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:** (516) _____ **Fax:** (516) _____

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

☐ **Peripheral Blood** ☐ **Products of Conception (POC)** ☐ **Skin Biopsy** ☐ **Tissue** _____

Date & Time Specimen Collected: ____/____/____ : ____

COMPLETE FOR PERIPHERAL BLOOD SPECIMENS**CLINICAL INDICATION**

- ☐ Autism ☐ Failure to thrive
☐ Developmental delay ☐ Intellectual disability
☐ Dysmorphism ☐ Recurrent pregnancy loss
☐ Rule Out Trisomy (specify:____) ☐ Other _____

TEST(S) REQUESTED

- ☐ **Chromosome Analysis (Karyotype)**
☐ **High Resolution Chromosome Analysis**
☐ **Mosaicism Study**

Fluorescence *in situ* Hybridization (FISH):

- ☐ **RAPID Newborn Aneuploidy FISH**
☐ Williams Syndrome (7q11.23)
☐ Prader-Willi/Angelman Syndrome (15q11.2)
☐ DiGeorge/Velo-Cardio-Facial Syndrome (22q11.2)
☐ Kallmann Syndrome (Xp22.3)
☐ SRY deletion (Yp11.3)
☐ **High Resolution Chromosome Microarray** (GeneDx 910)
Please include blood specimen from parents of the proband if available
(1 EDTA lavender top tube) ☐ Included ____ mother ____ father
☐ **Other:** _____

COMPLETE FOR POC/SKIN BIOPSY/TISSUE SPECIMENS**CLINICAL INDICATION**

- ☐ Missed abortion ☐ Fetal demise ☐ Molar pregnancy
☐ Neonatal demise ☐ Stillbirth
☐ Confirm prenatal analysis for: _____
☐ Other _____

For POCs/Neonatal Demise:

Weeks gestation _____

Tissue origin (please circle): Villi / Skin / Umbilical Cord / Other _____

TEST(S) REQUESTED

- ☐ **Chromosome Analysis (Karyotype)**
☐ reflex to HR microarray if no growth
☐ reflex to HR microarray if chromosomes are normal
☐ **High Resolution (HR) Chromosome Microarray** (GeneDx 460)
Please include blood specimen from parents of the proband if available
(1 EDTA lavender top tube) ☐ Included ____ mother ____ father
☐ **Hold cells for:** _____ ☐ **Grow cells for:** _____
☐ **Send out for:** _____ **to** _____
☐ **Other:** _____

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 and newborn aneuploidy 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.