Northwell Health® Genomics Alliance

PRENATAL GENETICS REQUISITION

OUTLINED AREAS MUST BE COMPLETED

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 _____

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	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: _____

ACCOUNT S4762 Northwell Health Genomics Alliance
T: (516) 719-1100 F: (516) 719-1220

PREGNANCY/PRECONCEPTION ASSESSMENT

ClariTest™ Non-Invasive Prenatal Screening (VERIFI)
(10 weeks+ /or singleton or twin pregnancies)

ClariTest cannot be performed on triplets or higher-order multiples. Microdeletions analysis cannot be performed for patients with a confirmed multiple gestation pregnancy.

☐ ClariNIPTM ClariTest w/microdeletions (2BCT)
☐ ClariNIPT ClariTest Only (2BCT)
Please include fetal gender: ☐ Y ☐ N
Is this a redraw? ☐ Y ☐ N

MOLECULAR GENETICS PROFILES

☐ Inherigen InheriGen Pan Ethnic Carrier Screen
☐ InherigenPLUS InheriGen Plus (includes InheriGen, CFPLUS, FRX and SMA)
☐ AshkJew 18 Ashkenazi Jewish 18 panel including CFPLUS
☐ Ashk Jew 25 Ashkenazi Jewish 25 Panel
☐ CFPLUS Expanded Cystic Fibrosis
☐ SMA Spinal Muscular Atrophy
☐ HLX Fragile X CASE Fragile X
☐ TAYP Tay Sachs Enzymes

CLINICAL INFORMATION

LAST MENSTRUAL PERIOD _____ EDD _____
☐ G _____ ☐ P _____ PATIENT WEIGHT _____ lbs
DATE OF ULTRASOUND _____
GA AT TIME OF ULTRASOUND _____ weeks _____ days
OF FETUSES _____ IF TWINS: ☐ Dichorionic ☐ Monochorionic
☐ IVF PREGNANCY _____ ☐ EGG Donor age retreival _____ ☐ SURROGATE
PREVIOUS PREGNANCY HISTORY OF
☐ ONTD ☐ DOWN SYNDROME ☐ GENETIC DISORDER ☐ OTHER _____
Gender/Karyotype (if known) _____

INDIVIDUAL MOLECULAR GENETICS

Ashkenazi Jewish 18 Profile

☐ BLMSYN Bloom Syndrome (L)
☐ CANADNA Canavan Disease (L)
☐ DIHYDEF Dihydrolipoamide Dehydrogenase Deficiency (L)
☐ CFPLUS Expanded CF Panel (>200 Mutations) (L)
☐ FDYS Familial Dysautonomia (L)
☐ FAMHYP Familial Hyperinsulinism (L)
☐ FAN Fanconi Anemia (Group C) (L)
☐ GAUDNA Gaucher Disease (L)
☐ GSD1A Glycogen Storage (Type 1A) (L)
☐ JOUBERT Joubert Syndrome (L)
☐ MAPLEDNA Maple Syrup Urine Diease (Type 1A & 1B) (L)
☐ MUCODNA Mucolipidosis (Type IV) (L)
☐ NEMALINE Nemaline Myopathy 2 (L)
☐ NEPK Niemann-Pick Disease (Type A & B) (L)
☐ TAY Tay-Sachs DNA (L)
☐ USHER1 Usher Syndrome Type 1F (L)
☐ USHER2 Usher Syndrome Type III (L)
☐ WALKER Walker Warburg Syndrome (L)

Expanded Ashkenazi Jewish Profile
(Includes Ashkenazi Jewish 18 Profile plus below tests)

8148-8 ☐ Abetalipoproteinemia (L)
J316-1 ☐ Alport Syndrome, Autosomal Recessive (L)
J297-3 ☐ Arthrogryposis, Mental Retardation & Seizures (L)
J317-9 ☐ Bardet-Biedl Syndrome 2 (L)
J315-3 ☐ Carnitine Palmitoyltransferase Deficiency, Type 2 (L)
J311-2 ☐ Ciliary Dyskinesia. Primary 1 (L)
J441-7 ☐ Coenzyme Q10 Deficiency, Primary, 7 (L)
J306-2 ☐ Cogenital Amegakaryocytic Thrombocytopenia (CAMT) (L)
J308-8 ☐ Deafness, Autosomal Recessive 1A (GJB2) (L)
J299-9 ☐ Dyskeratosis Congenita, Autosomal Recessive 5 (L)
J294-0 ☐ Ehlers-Danos Syndrome, Type VIIC (L)
J310-4 ☐ Factor XI Deficiency (Hemophilia C) (L)
B151-2 ☐ Familial Hypercholesterolemia LDLR Associated (L)
J307-0 ☐ Familial Mediterranean Fever (L)
B155-3 ☐ Galactosemia (L)
B150-4 ☐ Hermansky-Pudlak Syndrome 3 (L)
J305-4 ☐ Mitochondrial Complex 1 Deficiency (L)
J296-5 ☐ Multiple Sullatase Deficiency (L)
J29S-7 ☐ Osteopetrosis, Autosomal Recessive 1 (L)
J303-9 ☐ Peroxisome Biogenesis Disorder 5A (Zellweger) (L)
J304-7 ☐ Phenylketonuria (PKU) (L)
J302-1 ☐ Phosphoglycerate Dehydrogenase Deficiency (L)
J301-3 ☐ Polycystic Kidney Disease, Autosomal Recessive (ARPKD) (L)
J313-8 ☐ Retinitis Pigmentosa 59 (L)
J314-6 ☐ Smith-Lemli-Opitz Syndrome (L)
J298-1 ☐ Spastic Tetrapleglia, Thin Corpus Callosum & Progressive Microcephaly (L)
B149-1 ☐ Tyrosinemia, Type 1 (L)
B164-5 ☐ Wilson Disease (L)

INDICATIONS FOR TESTING

☐ Routine Screening ☐ Carrier Screening (No family History) ☐ Ashkenazi Jewish Ancestry ☐ AMA
☐ Abnormal Antenatal Screen for _____ ☐ Abnormal NIPT Result (include copy of report) ☐ Family History ☐ Previous Pregnancy History
☐ Clinical Suspicion of Disease _____ ☐ Infertility ☐ Recurrent Pregnancy Loss ☐ Other _____
☐ Abnormal Ultrasound Results (Please specify below)
☐ Cystic Hygroma: _____ ☐ Increased Nuchal Translucency/Nuchal Fold: _____ ☐ CNS: _____
☐ Uro-Genital: _____ ☐ Skeletal: _____ ☐ IUGR: _____
☐ Head/Neck: _____ ☐ Cardiac: _____
☐ Gastrointestinal: _____ ☐ Other: _____

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INFORMED CONSENT FOR GENETIC TESTING

Before signing this consent form, you should review the information below and discuss prenatal genetic testing with your healthcare provider or genetic counselor. By signing this consent, you authorize the collection of a suitable sample for genetic testing, you agree that you understand the following information, and you agree to the performance of the following genetic testing.

☐ Carrier Screening ☐ ClariTest

What is genetic testing?

DNA is the chemical that provides instructions for our body's growth and development. Genes are distinct sequences of DNA, which form part of a chromosome . The DNA sequence in a gene contains the instructions for making proteins, which in turn determine growth and metabolism as well as traits, such as eye color and blood type. Genetic disorders are caused by harmful changes in the sequence of a persons DNA or from changes in the structure or number of chromosomes. Genetic testing is a type of laboratory test that identifies these harmful changes in chromosomes or the DNA of a gene. The purpose of this test is to see if I, or my child, have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the cancer that I, or my child, will develop or pass on a genetic disorder in the future. 'My child' can also mean my unborn child for the purposes of this consent.

For genetic testing, a small specimen or blood, saliva or other suitable tissues will be collected and sent to the lab for DNA analysis. My results will be sent to the doctor that ordered the test and at my doctors request to any other healthcare provider or genetic counselor. It is recommended that I receive genetic counseling before and alter genetic testing I can find a genetic counselor in my area here: www.nsgc.org. Further testing or additional consultations with a healthcare provider may be necessary.

Test Information:

Additional information about the specific test being ordered is available from my health care provider or I can go to the GenPath website, www.genpathdiagnostics.com. This information includes the specific types of genetic disorders that can be identified by the genetic test, the likelihood or a positive result, and the limitations or genetic testing.

Carrier Screening is performed to determine if the chance is increased to have a baby w th an Inherited genetic disorder. Carriers of a genetic disorder are typically healthy individuals who do not show signs of the disease, yet possess a change in their genetic information, Most disorders screened for on carrier screening are Inherited in an autosomal recessive manner, which means that in both parents are carriers for the same disorder, there is a 25% chance of having a child with the disease. Cystic Fibrosis, Spinal Muscular Atrophy and most disorders on the InheriGen panel are inherited in an autosomal recessive manner. Fragile X syndrome and three disorders tested for on the InheriGen panel are inherited in an X-linked manner. In X-linked inheritance, if a woman is a carrier, there is up to a 50% chance of having a child with the disease. A negative, or normal, carrier screening result can reduce the chance that I am a carrier for any of the tested diseases but does not entirely eliminate this risk.

• **InheriGen** is a carrier screen that tests for disorders that are mostly severe, childhood onset diseases. InheriGen is a pan-ethnic carrier screen intended for the general population. Approximately 20 to 25% of individuals in the general population undergoing testing will be found to be a carrier for at least one condition. The detection rate for each disorder can vary by ethnicity, and more detailed information can be found on our website at www.genpath.com.

• **Cystic Fibrosis (CF)** is a disease that affects approximately 1/2500 to 3300 live births. It is a multisystem disease that affects the lungs, pancreas, gastrointestinal tract and reproductive systems . Approximately 1/25 Caucasians, 1/60 African Americans or Hispanics, and 1/90 Asians are earners for CF. CF can be screened for by two different panels, a panel with 40 pathogenic variants or the Expanded CF Panel, which tests for 215, pathogenic variants . The detection rates are generally greater for most ethnicities when the 215 pathogenic variant panel is used.

• **Spinal Muscular Atrophy (SMA)** is the most common inherited lethal disease in children; involving the degeneration of cells that control movement in the spinal cord, leading to muscle weakness and wasting. The earner frequency is between 1/25 and 1/50 in the general population . Detection rates vary by ethnicity and are available on our website at www genpath.com.

Noninvasive Prenatal Testing (NIPT): This test screens for specific chromosomal abnormalities in a pregnancy by looking at the DNA in the mother's blood . To determine whether too few or too many chromosomes are present in a fetus, this test uses a technology called 'massively parallel DNA sequencing' to count the number of copies of the specific chromosomes, and then uses a proprietary method to determine if there are too many or too few copies of the chromosomes in the pregnancy.

• **ClariTest** is a NIPT test that screens for the presence of trisomy 21 (Down syndrome), trisomy 18 (Edward syndrome), trisomy 13 (Palau syndrome) as well as the sex chromosomes (X and Y), It can also test for microdeletions of certain chromosomes including 22q11.2 deletion (DiGeorge), 15q11 deletion (Angelman/Prader·Willi), 1p36 deletion, 4p- (Wolf-Hirschhorn), and 5p- (Cri-du-chat). This test can be performed starting at 10 weeks of pregnancy. This is a screening test that only looks for specific chromosomal abnormalities in a fetus. This test does not test the health of the mother. Normal test results do not eliminate the possibility that your pregnancy may have other chromosomal abnormalities, birth defects, or other conditions, such as open neural tube defects. In addition, a normal result does not guarantee a healthy pregnancy or baby . This test has limitations including the possibility of false positive and false negative results. This test can reduce, but not eliminate the risk for specific chromosome abnormalities and microdeletions. GenPath recommends that no irreversible clinical decisions be made based on these screening results alone . If definitive diagnosis is desired, chorionic viflous sampling or amniocentesis would be necessary. As explained above, genetic counseling before and after testing is recommended.

By signing this form I agree and understand that:

- *A sample of blood may be drawn by venipuncture, a procedure which carries a negligible risk.*
- *No testing other than those marked on the reverse of this form will be performed on this sample.*
- *I have the option to obtain genetic counseling before signing the informed consent.*
- *The laboratory may retain any remaining non-NY sample after completion of testing and use my de-identified sample for internal validation, quality control and quality assurance.*
- *The state of New York has a rule that specimens from New York residents cannot be retained by the laboratory longer than 60 days after test completion unless it is authorized by the patient NY residents. ONA sample can be de-identified and retained for greater than 60 days after the completion of testing for use as quality control material. (Please initial to consent) _____*
- *If I am a carrier of any of the above diseases, the probability of detecting a pathogenic variant by molecular methods can be dependent on my ethnicity and specific variants analyzed. Information on the detection rate for each individual disease based on ethnicity is available upon request and will be included in the results report. If I am positive for any of the molecular genetic tests (i.e I am a carrier) genetic counseling will be recommended to me and additional recommendations for testing may be provided.*
- *My carrier screening and NIPT result, when negative, only applies to variants and disorders analyzed. There remains a chance that I may be a carrier of a pathogenic variant or a pregnancy is affected with another disorder that was not part of this test, as these tests cannot detect all mutations/disorders.*
- *NIPT testing is a screening test and that diagnostic testing is recommended fo/lowing a positive result.*
- *In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering th is test. For example, this test may tell me about the risk for another genetic condition. This information may be disclosed ta the ordering healthcare provider if it likely impacts medical care.*
- *In rare instances, it may be necessary to obtain another sample in order to determine a result. This will be at no additional cost, from GenPath*
- *The methods used by GenPath are highly accurate ; however, rare errors can occur due to mislabeling of samples, bone marrow transplantation, blood transfusion, incorrect reporting of family history or relationships, or because some abnormalities are present In such a small traction of cells that they may not be detectable (mosaicism) ,*
- *Results will be reported to the indicated healthcare provider and if noted, copied to the additional healthcare provider Indicated on the front of this form . I understand that results may only be disclosed to others by my written consent and/or if demanded by an order of a court of competent jurisdiction.*
- *DNA specimens will not be returned to me or my healthcare provider unless specific prior arrangements have been made.*
- *I hereby authorize the laboratory to furnish my designated insurance carrier the information on this form if necessary for reimbursement. I also authorize payment to the laboratory. I understand that I am responsible for any amount not paid by insurance for reasons including, but not limited to, non-covered and non-authorized services. I permit a copy of this authorization to be used in place of the original.*

Patient Name: _____ Date of Birth: _____

(Please print) First Name Middle Name Last Name mm/dd/yyyy

Patient Signature: _____ Date: _____

Note for Healthcare Provider: It is the physician's responsibility to obtain informed consent from the patient/guardian. If a signed consent form is not forwarded to the laboratory, it is believed that the physician has obtained con sent and that the patient's signature is on life in his/her medical records.

Note: Patient consent is required In the following states : Alaska, Arizona, Florida, Georgia, Massachusetts, Michigan, Nebraska, New Mexico, New York, South Carolina, South Dakota, and Vermont. Patient consent is suggested in all other states .

The New York State Civil Rights Act, Section 79·1 requires that all individuals be informed of the nature of the genetic testing being requested.