

MOLECULAR (DNA) TEST REQUISITION

FOR CHG LAB USE ONLY:

Pedigree #: _____ Lab #: _____

Patient Name: _____
Last First MI

Date of Birth: _____

****SPECIMEN REQUIREMENTS FOR ALL TESTS LISTED BELOW: 7-10 cc BLOOD IN EDTA OR ACD ANTICOAGULANT****

DNA TEST(S) REQUESTED:

(PLEASE NOTE: ANALYSIS = SEQUENCING AND MLPA)

Date Sample Collected: _____

- 46,XY Disorders of sex development (**NR5A1** analysis)
- Aarskog Scott syndrome (**FGD1** analysis)
- Acute Myeloid (Myelogenous) Leukemia
 - FLT3** C835 mutation **NPM1** exon 12 sequencing
- Alpha-thalassemia/XLID syndrome (**ATRX** sequencing)
- Angelman/Angelman-like syndrome panel
 - Methylation-sensitive MLPA **UBE3A** sequencing
 - SLC9A6** sequencing **TCF4** analysis **ZEB2** analysis
- Ashkenazi Jewish panel
 - Bloom syndrome Gaucher disease
 - Canavan disease GSD Type 1A
 - Cystic fibrosis MSUD Type 1B
 - Factor XI deficiency Mucopolidosis type IV
 - Familial Dysautonomia Niemann-Pick disease Type A
 - Fanconi anemia Group C Tay Sachs disease
- Ataxia panel
 - Spinocerebellar ataxia (circle tests) Type 10, 12, 17
 - DRPLA
- Autism (with macrocephaly)
 - (**PTEN** analysis and promoter sequencing)
- Banking (circle type) DNA or Lymphoblast
- BCR/ABL
- Beals syndrome (**FBN2** sequencing)
- Borjesen-Forsman-Lehmann syndrome (**PHF6** sequencing)
- Branchio-oculo-facial syndrome (**TFAP2A** analysis)
- BRCA1/2 (Ashkenazi Jewish mutations only)**
- Cardiofaciocutaneous syndrome panel
 - Sequencing of **BRAF** **MAP2K1** **MAP2K2** **KRAS**
- Charcot-Marie-Tooth disease (Types 1B, 2I, 2J)
 - (**MPZ** sequencing)
- CHARGE syndrome (**CHD7** analysis)
- Coffin-Lowry syndrome (**RSK2** analysis)
- Colon Cancer
 - Familial Adenomatous Polyposis (**APC** analysis)
 - Lynch/HNPCC: (**MLH1**, **MSH2**, **MSH6**, & **PMS2** analyses; **EPCAM/TACSTD1** MLPA)
 - MYH** Associated Polyposis (**MUTYH** analysis)
- Congenital bilateral absence of vas deferens (CBAVD)
 - CF 100 mutation panel **CFTR** analysis
- Congenital Contractural Arachnodactyly (**FBN2** sequencing)
- Connexin 30 deletion (nonsyndromic deafness)
- Costello syndrome panel
 - Sequencing of **HRAS** **KRAS** **BRAF**
- Cystic fibrosis
 - 40 mutation panel 100 mutation panel
 - CFTR** analysis
- Ehlers-Danlos syndrome Type IV (**COL3A1** analysis)
- Factor V Leiden
- Familial Mediterranean Fever
 - Common mutations only **MEFV** analysis
- Fragile X syndrome
- Gene expression studies (RT-PCR) _____
- Hemochromatosis
- Huntington disease**
- Infertility testing
 - Ovarian insufficiency/spermatogenic failure (**NR5A1** analysis)
 - Premature ovarian failure (Fragile X testing)
 - SYCP3** sequencing
 - Y-microdeletion studies
- JAK2 (V617F mutation [reflex to exon 12 sequencing])
- Kabuki syndrome (**MLL2** analysis)
- Kennedy disease (SBMA)
- LADD syndrome (**FGF10** analysis)
 - (reflex to **FGFR2** analysis and **FGFR3** sequencing)
- LEOPARD syndrome panel
 - Sequencing of **PTPN11** **RAF1** **BRAF**
- Loey's-Dietz syndrome
 - Analysis of **TGFBR1** **TGFBR2**
- Marfan syndrome (**FBN1** analysis)
- Maternal cell contamination studies
- MCAD
- MED12** related disorders (**MED12** sequencing)
- Melanoma (**CDKN2A** analysis)
- Mitochondrial diseases panel
 - All 37 gene sequencing MELAS
 - CPEO/KSS LHON
 - Leigh syndrome/NARP MERRF
- Mowat Wilson syndrome (**ZEB2** analysis)
- MTHFR
- Multiple Endocrine Neoplasia, type 1 (**MEN1** analysis)
- Multiple Endocrine Neoplasia, type 2 (**RET** analysis)
- Neurofibromatosis
 - Analysis of **NF1** **NF2**
- Neurofibromatosis-Noonan syndrome (**NF1** analysis)
- Neurofibromatosis type 1-like (Legius) syndrome
 - (**SPRED1** analysis)
- Neuroligin (**NLGN3** and **NLGN4** sequencing)
- Noonan syndrome panel
 - Sequencing of **PTPN11** **SOS1** **RAF1**
 - KRAS** **NRAS** **SHOC2** S2G mutation
- OpitzG/BBB syndrome (**MID1** analysis)
- Pancreatitis (hereditary)
 - Pancreatitis panel: **CTRC** sequencing, Analysis of **PRSS1**, **SPINK1**
 - CFTR** analysis
- Paraganglioma-Pheochromocytoma syndromes
 - Analysis of **SDHB** **SDHC** **SDHD**
- Paternity testing (Call before sending samples)
- Pelizaeus-Merzbacher disease (**PLP1** analysis)
- Pendred syndrome
- Phenylketonuria (**PAH** analysis)
- Pitt Hopkins syndrome (**TCF4** analysis)
- Prader-Willi syndrome
- Prothrombin (G20210A)
- PTCHD1** analysis
- PTEN** Hamartoma Tumor syndromes
 - (**PTEN** analysis and promoter sequencing)
- Renpenning syndrome (**PQBP1** analysis)
- Rett/Rett-like syndrome panel
 - Analysis of **MECP2** **CDKL5** **FOXG1** **TCF4**
- Sickle cell anemia (HbSS, HbSC)
- Smith-Lemli-Opitz syndrome (**DHCR7** sequencing)
- SNP microarray
- Sotos syndrome (**NSD1** analysis)
- Stickler syndrome Type 1 (**COL2A1** analysis)
- SYNGAP1** sequencing
 - (Intellectual disability, dominant, nonsyndromic)
- Tay-Sachs disease
- Thoracic Aortic Aneurysms/Dissections panel
 - MYH11** sequencing **TGFBR1** analysis
 - TGFBR2** analysis **FBN1** analysis
 - SMAD3** sequencing **ACTA2** sequencing
- Thrombophilia panel (Factor V Leiden, Prothrombin, MTHFR)
- Tuberous Sclerosis (**TSC1** analysis)
- UPD: (circle test) chromosome 7 14 15
- von-Hippel-Lindau disease (**VHL** analysis)
- Waardenburg syndrome
 - Types 1 and 3: **PAX3** analysis
 - Type 2: Analysis of **MITF**, **SOX10**
 - Type 4: Analysis of **SOX10**, **EDN3**, **EDNRB**
- Wilson disease (**ATP7B** analysis)
- X-inactivation studies
- X-linked lymphoproliferative disease (**SH2D1A** analysis)
- X-linked Intellectual Disability (XLID)
 - (Order whole panel, individual tiers, or single gene)
 - Tier A **NLGN3** sequencing **NLGN4** sequencing
 - Rett syndrome (**MECP2** analysis)
 - Rett syndrome - atypical (**CDKL5** analysis)
 - Tier 1 **DLG3** sequencing **FTSJ1** sequencing **JARID1C** sequencing
 - Borjesen-Forsman-Lehmann syndrome (**PHF6** sequencing)
 - ZNF41** sequencing
 - Tier 2 Asperger syndrome (**GDI1** analysis) **FACL4** analysis **OPHN1** analysis Renpenning syndrome (**PQBP1** analysis) **TM4SF2/ITSPAN7** analysis
 - Tier 3 Alpha-thalassemia/XLID syndrome (**ATRX** sequencing) Aarskog Scott syndrome (**FGD1** analysis) OpitzG/BBB syndrome (**MID1** analysis) Pelizaeus-Merzbacher disease (**PLP1** analysis) Coffin-Lowry syndrome (**RSK2** analysis) Creatine (transporter) deficiency (**SLC6A8** analysis)
 - Tier 4 **AGTR2** analysis **ARHGEF6** analysis **MED12** sequencing **PAK3** analysis **SLC16A2** sequencing
- Other X-linked intellectual disability genes
 - RAB39B** sequencing
- X-linked Intellectual Disability/Epilepsy Panels
 - Panel 1 Angelman-like syndrome (X-linked Christianson type) (**SLC9A6** sequencing) **PCDH19** sequencing (females only) Rett syndrome (**MECP2** analysis) Rett syndrome - atypical (**CDKL5** analysis)
 - Panel 2 **ATP6AP2** sequencing Creatine (transporter) deficiency (**SLC6A8** analysis) **OPHN1** analysis **SYN1** sequencing
- Y-microdeletion studies/Y-chromosome detection (SRY)
- Zygosity testing

*****BILLING INFORMATION AND COPY OF INSURANCE CARD FRONT
AND BACK MUST ACCOMPANY SAMPLE AND REQUISITION FORM*****

SVC PROVIDER: CENTER FOR HUMAN GENETICS INC@BUSM CLIA #22D0650242 NPI #1821153156

PATIENT INFORMATION:

LAST NAME:	GENDER: (CIRCLE)	DATE OF BIRTH
FIRST NAME:	M F	 / /
MIDDLE:		MM/DD/YYYY
STREET ADDRESS:		APARTMENT# / FLOOR
CITY :	STATE	ZIP
PHONE: HOME()	CELL()	

PAYMENT INFO: (SELECT ONE) (CIRCLE): LAB/HOSP/FAC/INST INSURANCE PATIENT CREDIT CARD

BILLING INFORMATION (MUST BE COMPLETED)

LABORATORY/ HOSPITAL/ FACILITY/ INSTITUTIONAL BILLING ADDRESS:

FACILITY NAME:	AFFIX LABEL HERE:
ADDRESS:	
CITY, STATE, ZIP	
ATTENTION:	
PHONE: ()	
FAX: ()	
PURCHASE ORDER#	
PATIENT MEDICAL RECORD#	

INSURANCE INFORMATION:

INSURANCE COMPANY NAME:
INSURANCE IDENTIFICATION #
INSURANCE GROUP #

SUBSCRIBER NAME:	SUBSCRIBER DATE OF BIRTH:
LAST:	 / /
FIRST:	MM / DD / YYYY

RELATIONSHIP TO PATIENT:	(CIRCLE):
	SELF PARENT SPOUSE

INSURANCE ADDRESS:	INSURANCE TELEPHONE AND EXTENTION:
STREET:	()
	FAX# ()
	CONTACT NAME/DEPT:

CITY, STATE, ZIP:	AUTHORIZATION#
	VALID FROM: / / TO / /

*SECONDARY INS NAME:	SUB NAME:
RELATIONSHIP:	
POLICY #	SUB DOB: / /
GENDER: M F	

PATIENT ACKNOWLEDGEMENT:
I AUTHORIZE ANY HOLDER OF MEDICAL INFORMATION ABOUT ME TO RELEASE TO ANY INSURANCE CARRIER ANY INFORMATION NEEDED FOR THIS CLAIM. I PERMIT A COPY OF THIS AUTHORIZATION TO BE USED IN PLACE OF THE ORIGINAL AND REQUEST THAT THE PAYMENT OF MEDICAL INSURANCE BE PAID TO CHG, INC. I ALSO UNDERSTAND THAT I WILL BE HELD RESPONSIBLE FOR ANY PORTION OF THE CLAIM THAT THE INSURANCE COMPANY DOES NOT PAY.

REQUIRED SIGNATURE:
DATE: / /

MEDICARE ID#: **BENEFICIARY NAME:**

BENEFICIARY AGREEMENT:
I HAVE BEEN NOTIFIED BY THE CENTER FOR HUMAN GENETICS THAT, IN MY CASE, MEDICARE IS LIKELY TO DENY PAYMENT FOR THE SERVICES IDENTIFIED BELOW, FOR THE REASON STATED. IF MEDICARE DENIES PAYMENT, I AGREE TO BE PERSONALLY AND FULLY RESPONSIBLE FOR PAYMENT.

REQUIRED BENEFICIARY SIGNATURE:
DATE: / /

MEDICARE WILL ONLY PAY FOR SERVICES THAT IT DETERMINES TO BE "REASONABLE AND NECESSARY" UNDER SECTION 1862(a) (1) OF THE MEDICARE LAW.
IF MEDICARE DETERMINES THAT A PARTICULAR SERVICE, ALTHOUGH IT WOULD OTHERWISE BE COVERED, IS NOT "REASONABLE AND NECESSARY" UNDER MEDICARE PAYMENT STANDARDS, MEDICARE WILL DENY PAYMENT FOR THAT SERVICE. THE CENTER FOR HUMAN GENETICS BELIEVES THAT MEDICARE IS LIKELY TO DENY PAYMENT FOR MOLECULAR DNA TESTING.
PROVIDER #228243

MOLECULAR (DNA) TEST REQUISITION

Center for Human Genetics, Inc.

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Phone: (617) 638-7083 Fax: (617) 638-7092

Web: <http://www.bumc.bu.edu/hg>

Patient Name: _____
Last First MI

Date of Birth: _____

INFORMED CONSENT FOR DNA TESTING

**** A signed consent form is REQUIRED for all samples from New York and all predictive testing BEFORE testing can be initiated.
** A signed consent form is strongly recommended for all genetic testing. If a signed consent is not submitted, the Center for Human Genetics assumes that the ordering physician has appropriately reviewed and obtained the patient's informed consent**

I/We request and authorize the DNA Diagnostic Laboratory at the Center for Human Genetics to analyze a sample of DNA isolated from _____ (sample type) obtained on _____ (date) to assess the probability that I (my/our fetus/child) am (is) affected with or carry the gene for the genetic disease _____ which is _____.

The test procedure has been explained to me/us and I/we understand that:

- I. There are several possible outcomes of this test:
 - 1. The test results may indicate that it is likely or unlikely that I (my/our fetus/child) am (is) affected with, or a carrier for, the above disease.
 - 2. The test results may be indeterminate because of my (my/our fetus/child's) genetic patterns or the genetic patterns of my family members (if also tested), and/or the limitations of the current technology.
- II. DNA tests are performed with precision and results reflect great accuracy and specific degrees of quoted accuracy (when applicable). Turn-around time is estimated and cannot be guaranteed.
- III. One possible result of DNA testing is that the laboratory could discover evidence of previously undisclosed non-paternity when comparing my (my/our fetus'/child's) sample with samples from other family members.
- IV. Genetic counseling, further testing, or additional physician consults may be warranted after testing in order to complete the testing process.
- V. After the DNA testing of my (my/our fetus'/child's) sample is complete, DNA will be stored at the Center for Human Genetics for a minimum of three months. We do not guarantee the future availability of DNA unless specific arrangements have been made for DNA banking. After that time, any remaining material will be disposed of at the discretion of the Laboratory Director, and may be used for medical research or education so long as our privacy is maintained.

I can request that remaining DNA not be used for research purposes by initialing here: _____

VI. The results of this test are to be released only to the ordering physician and referral laboratory (if applicable) per HIPAA regulations.

My/our signature(s) below constitute(s) my/our acknowledgement (1) that the proposed DNA test(s) and its/their limitations for my/our specific situation have been satisfactorily explained to me/us by my/our physician or genetic counselor; and (2) I/we hereby give my/our authorization and consent for this testing.

Patient/Guardian Signature

Witness Signature

Date