DNA Diagnostic Lab at JOHNS HOPKINS
Test Requisition Part I of II

Shipping Address:
1812 Ashland Ave
Sample Intake Rm 245
Baltimore. MD 21205

Referrer Information

Physician	UPIN /NPI
Genetic Counselor	Contact Email:
Institution	
Address City/State/Zip	
Phone	Fax

Additional Reports to

Name /Institution	
Address City/State/Zip	
Phone	Fax

Patient Information

*Two of these identifiers must appear on the sample

*Name (Last)		(First)	
Address City/State/Zip			
*Date of Birth (mm/dd/yyyy)	Sex	Ethnicity	Position in Pedigree
*Patient ID / Sample Number			

Sample Type **Date Collected: _____**

☐ Venous Blood ☐ Cord Blood ☐ Cleaned Chorionic Villi
☐ Cultured Amniocytes ☐ Cultured Chorionic Villi ☐ Cultured Fibroblasts ☐ Other Culture _____
☐ Frozen tissue (source: _____) ☐ DNA* (source: _____)
☐ PureGene Extraction ☐ Other method _____

*The DNA Diagnostic Laboratory only accepts isolated or extracted nucleic acids for which the extraction or isolation is performed in a CLIA-certified laboratory or a laboratory meeting equivalent (or more stringent) requirements as determined by the College of American Pathologists (CAP) and/or the Centers for Medicare and Medicaid Services (CMS)

Reason for Test ☐ Prenatal ☐ Carrier ☐ Targeted Variant Identification ☐ Presymptomatic ☐ Family member for Linkage analysis ☐ Confirmatory/Diagnostic

Diagnosis Code (ICD-10): _____ If the patient is pregnant: LMP _____

Billing Information

(All required documents must be received for testing to be initiated; see web site billing page for information)

☐ Credit Card – Attach Credit Card Authorization Form

☐ Check (Check # _____ Amount of Check: $ _____)
☐ Patient Insurance - **Contact Billing Coordinator at 443-287-2486 prior to submitting.**
☐ Referring Center (Include address and contact person if different from that provided above)
☐ Maryland Medicaid # _____ (referral required)
☐ Medicare # _____ (waiver form required)

For Internal/DNA Diagnostic Lab Use Only			
Accession #	Date Received	ID #	
Test 1:	Fee 1:	Test 2:	Fee 2:
Notes:		☐ Fax number(s) listed confirmed	

Phone: 410-955-0483 Fax: 410-955-0484 *www.hopkinsmedicine.org/dnadiagnostic/* ddl@jhmi.edu

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Test Requisition Part II of II

Shipping Address:
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Sample Intake Rm 245
Baltimore. MD 21205

Patient Name: _____ Patient DOB: _____

Panel / Test Requested (see website for details) **Check here** ☐ **if ordering a targeted test for a known variant***

Multi-Gene Panels / Next Generation Sequencing (NGS)
(NGS panels include sequencing and deletion/duplication analysis of most genes; see website for exceptions)

☐ **Ataxia telangiectasia (*ATM*) NGS**

Cystic fibrosis and Related Disorders
☐ Cystic fibrosis (*CFTR)* NGS
☐ *CFTR* and CF-related disorders NGS panel (with *CFTR*)
☐ CF-related disorders NGS panel (without *CFTR*)

Peroxisomal Disorders
☐ Acatalasemia (*CAT*) NGS
☐ Alpha-Methylacyl-CoA racemase deficiency (*AMACR*) NGS
☐ Comprehensive peroxisomal genes NGS panel
☐ Mulibrey nanism (*TRIM37*) NGS
☐ Peroxisomal ß-oxidation defects NGS panel
☐ Primary hyperoxaluria type I (*AGXT*) NGS
☐ Refsum disease NGS panel
☐ Rhizomelic chondrodysplasia punctata NGS panel
☐ Zellweger spectrum disorders (ZSD) NGS panel
☐ ZSD + peroxisomal ß-oxidation defects NGS panel

☐ **Pseudohypoaldosteronism (PHA) type 1 NGS panel**

Surfactant disorders / Diffuse lung diseases
☐ Childhood interstitial lung disease NGS panel
☐ Comprehensive diffuse lung disease NGS panel
☐ Idiopathic pulmonary fibrosis NGS panel
☐ Neonatal respiratory distress NGS panel
☐ Pulmonary alveolar proteinosis NGS panel

☐ **Telomere Shortening Disorders Spectrum NGS Panel**
Please select proband's phenotype:
☐ Aplastic anemia - Myelodysplastic syndrome
☐ Coats plus syndrome
☐ Dyskeratosis congenita
☐ Hoyeraal-Hreidarsson syndrome
☐ Pulmonary fibrosis
☐ Revesz syndrome

☐ **Treacher Collins syndrome NGS panel**

Non-NGS Tests

Craniofacial Disorders
☐ Apert syndrome (*FGFR2*)
☐ Antley Bixler-like syndrome (*FGFR2*)
☐ Crouzon syndrome (FGFR2, 3)
☐ Jackson-Weiss syndrome (*FGFR2*, 3)
☐ Pfeiffer syndrome (*FGFR1*, 2, 3)
☐ Coronal synostosis (*FGFR2*, 3)
☐ Saethre-Chotzen syndrome (FGFR2, 3; *TWIST1* (sequencing; reflex to *TWIST1* MLPA)
☐ Crouzon with acanthosis nigricans (*FGFR3*)

☐ Craniofrontonasal syndrome (*EFNB1*)
☐ Oculodentodigital dysplasia (*GJA1*)

Endocrine Disorders
GNAS Spectrum
☐ Pseudohypoparathyroidism type 1A (PHP1A) / Albright hereditary osteodystrophy
(*GNAS* sequence, reflex to *GNAS* e1A methylation)
☐ Pseudohypoparathyroidism type 1b (PHP1B)
(*GNAS* exon *1A* methylation)

Familial Aortic Aneurysm Syndromes
☐ Loeys-Dietz syndrome (*TGFBR2 / TGFBR1*)
☐ Loeys-Dietz syndrome Type 1C (*SMAD3*)
☐ Marfan syndrome (Type 2; *TGFBR2*)

Skeletal Dysplasias
☐ Achondroplasia / Hypochondroplasia (*FGFR3*)
☐ Achondroplasia / Thanatophoric dysplasia (*FGFR3*)
☐ SADDAN (*FGFR3*)
☐ Campomelic dysplasia (*SOX9*)

REMINDER: MLPA testing should be performed on blood samples or PureGene extracted DNA samples only. Be sure to note the extraction method on P.1.

Huntington Disease Phenotype
☐ Huntington disease (*HTT* repeat sizing)
☐ Huntington disease like 2 (*JPH3* repeat sizing)

Other Conditions
☐ Benign hereditary chorea (*NKX2-1* sequence; reflex to MLPA)
☐ *CFTR* Intron 8 T/TG tract only
☐ Choreoathetosis, hypothyroidism, respiratory distress syndrome (*NKX2-1* sequence; reflex to MLPA)
☐ Hereditary non-syndromic sensorineural hearing loss (*GJB2* sequence; *GJB6* deletion assay)
☐ Liddle syndrome (*SCNN1B* and *SCNN1G*)
☐ von Hippel-Lindau syndrome (*VHL*)
  ☐ *VHL* sequence; reflex to MLPA
  ☐ *VHL* sequence plus MLPA
☐ X-Adrenoleukodystrophy (*ABCD1* sequence; reflex to MLPA)
  ☐ Copy result to Dr. Ali Fatemi (KKI)

Linkage Assays
☐ Cystic fibrosis (*CFTR*) linkage analysis
☐ Duchenne/Becker muscular dystrophy (*DMD*) linkage analysis
* All linkage analysis testing must be coordinated with the lab genetic counselor prior to submitting

☐ Maternal cell contamination study

Notes/Clinical Information:

***Targeted variant testing and prenatal testing for any of the above conditions **must** be arranged with the lab prior to shipping. Call 410-955-0483.

Informed Consent:
I understand that my physician is requesting the DNA Diagnostic Lab of the Johns Hopkins School of Medicine to perform the genetic test selected above on me / my child. The purpose and accuracy of this testing have been reviewed by my health care provider and my questions about these issues have been answered. In some cases it is necessary to do an indirect test that does not identify a specific disease causing variant. If I am to have an indirect test, my health care provider has discussed these issues with me. I understand that in most cases, a negative test result does not necessarily rule out a genetic condition. Results of genetic testing should be considered with the results of other types of testing and clinical evaluation. Lack of cooperation of all needed family members may compromise the quality or decrease the accuracy of the result obtained. If multiple family members are being tested, non-paternity may be disclosed by these results. No clinical tests other than those authorized will be performed; however, any remaining sample may be used quality control purposes or research after de-identification. The laboratory cannot guarantee turn-around time or that a result will be obtained on any sample. Results will be released only to parties indicated on the test requisition or their agents. Release to other parties requires written consent of the patient.

Signed: _____ Date: _____

Alternate Consent:
I, the health care provider requesting the above testing, have explained the benefits and drawbacks of genetic testing to the patient and have obtained verbal consent or an alternate written consent (please attach) to order the above test.

Signed: _____ Date: _____

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