



Requisition Form

The University of Chicago Genetic Services Laboratories

5841 South Maryland Avenue, Room G701/MC0077, Chicago, IL 60637

Toll Free: 888.824.3637 | Local: 773.834.0555 | Fax: 773.702.9130

ucgslabs@genetics.uchicago.edu | dnatesting.uchicago.edu | CLIA#: 14D0917593 | CAP#: 18827-49

Patient Information

Name: Last _____ First _____ Date of Birth (mm/dd/yyyy): _____

Gender: ☐ Male ☐ Female MRN: _____

Ethnicity: ☐ Caucasian ☐ African-American ☐ Hispanic ☐ Asian ☐ Ashkenazi Jewish ☐ Other _____

Ordering Physician Information

REPORTING RESULTS: Reports will only be faxed out. Please check the boxes below for those who should receive by fax.

☐ Referring Physician: _____

Phone: _____ Fax: _____

Email: _____

☐ Genetic Counselor: _____

Phone: _____ Fax: _____

Email: _____

☐ Referring Lab: _____

Phone: _____ Fax: _____

Email: _____

Indication for Testing

REQUIRED INFORMATION. NECESSARY FOR TESTING

☐ Symptomatic: _____ ICD-10: _____

Results of previous genetic testing: _____

☐ Asymptomatic/Positive Family History: (Mutation unknown – Please provide family history) Relationship to Proband: _____

☐ Testing for known mutation/variant*: Gene Name: _____ Mutation/Variant: _____

☐ Symptomatic ☐ Asymptomatic Name of Proband/UoC Lab Number: _____ Relationship to Proband: _____

☐ Other (Please specify clinical findings below): _____

*Requires prior approval by UCGS Lab Staff if this is a gene for which we do not offer full sequencing.

Sample Information

Date Sample Drawn (mm/dd/yyyy): _____

Specimen Type: ☐ Peripheral Blood (EDTA tube) ☐ Peripheral Blood (NaHep tube – for SNP array only) ☐ Peripheral Blood (PAX tube) ☐ Amniotic Fluid
☐ Chorionic Villi ☐ POC ☐ Saliva ☐ Buccal ☐ DNA (please specify original sample type: _____) ☐ Culture: _____

For prenatal specimens, please indicate current gestational age: _____ weeks by: ☐ LMP ☐ Ultrasound

Specimen Requirements: Routine Tests: 3-10cc blood in an EDTA (purple top) tube (unless otherwise indicated).

Prenatal Tests: 5-7cc amniotic fluid, 25-30mgs chorionic villi or 2 T25 flasks of cultured cells. Note, if direct amniotic fluid or chorionic villi are being sent, please start a back-up culture at your institution. Please also send 3-10cc of mother's blood in an EDTA tube for maternal cell contamination studies.

The sensitivity of our deletion/duplication and next generation sequencing assays may be reduced when an outside laboratory extracts DNA. For best results, please provide a fresh blood sample for these tests.

Note: All samples should be shipped via overnight delivery at room temperature to the address at the top of this page. No weekend or holiday deliveries. Label each specimen with the patient's name, date of birth and date sample collected.

Ordering Checklist

- ☐ Test Requisition Form (required)
- ☐ Completed Indication for Testing/ICD-10 study code (required)
- ☐ Completed Billing Information (required)
- ☐ Completed Research Consent Form (recommended)

For Office Use Only

TEST REQUESTS - Requisition Form

The University of Chicago Genetic Services Laboratories

Angelman syndrome testing

- ☐ Methylation Specific-MLPA
- ☐ UPD15 (requires samples from both parents also)
- ☐ Imprinting Center Deletion Analysis
- ☐ Angelman Syndrome Tier 2 Panel
- ☐ Rett/Angelman Syndrome Sequencing Panel
- ☐ Rett/Angelman Syndrome Deletion/Duplication Panel
- ☐ UBE3A Sequencing ☐ UBE3A Del/Dup
- ☐ SLC9A6 Sequencing ☐ SLC9A6 Del/Dup

Brain malformation testing

Cerebellar/Pontocerebellar Hypoplasia (PCH) testing

- ☐ Cerebellar/Pontocerebellar Hypoplasia Sequencing Panel
- ☐ Cerebellar/Pontocerebellar Hypoplasia Deletion/Duplication Panel
- ☐ TSEN54 Sequencing ☐ TSEN54 Del/Dup
- ☐ CASK Sequencing ☐ CASK Del/Dup
- ☐ OPHN1 Sequencing ☐ OPHN1 Del/Dup

Cerebral Cortical Malformation testing

- ☐ Cerebral Cortical Malformation Sequencing Panel
- ☐ Cerebral Cortical Malformation Deletion/Duplication Panel

Holoprosencephaly testing

- ☐ Holoprosencephaly Sequencing Panel
- ☐ Holoprosencephaly Deletion/Duplication Panel

Hydrocephalus testing

- ☐ Comprehensive Hydrocephalus Panel
- ☐ L1CAM Sequencing ☐ L1CAM Del/Dup
- ☐ Autosomal Recessive Non-Syndromic Hydrocephalus Sequencing Panel
- ☐ Autosomal Recessive Non-Syndromic Hydrocephalus Deletion/Duplication Panel

Lissencephaly testing

- ☐ Comprehensive Lissencephaly Panel
- ☐ Lissencephaly Sequencing Panel
- ☐ Lissencephaly Deletion/Duplication Panel
- ☐ Cobblestone Lissencephaly Sequencing Panel
- ☐ Cobblestone Lissencephaly Deletion/Duplication Panel
- ☐ DCX Sequencing ☐ DCX Del/Dup
- ☐ PAFAH1B1 (LIS1) Sequencing ☐ PAFAH1B1 (LIS1) Del/Dup
- ☐ TUBA1A Sequencing ☐ TUBA1A Del/Dup
- ☐ ARX Sequencing ☐ ARX Del/Dup

Polymicrogyria testing

- ☐ Polymicrogyria Sequencing Panel
- ☐ Polymicrogyria Deletion/Duplication Panel
- ☐ GPR56 Sequencing ☐ GPR56 Del/Dup
- ☐ OCLN Sequencing (Exons 2-5 only) ☐ OCLN Del/Dup (Exons 2-5 only)
- ☐ TUBB2B Sequencing ☐ TUBB2B Del/Dup
- ☐ TUBB3 Sequencing ☐ TUBB3 Del/Dup

Chondrodysplasia punctata testing

- ☐ ARSE Sequencing ☐ ARSE Del/Dup
- ☐ EBP Sequencing ☐ EBP Del/Dup
- ☐ Rhizomelic Chondrodysplasia Punctata Sequencing Panel

Ciliopathy Testing

- ☐ Bardet Biedl Syndrome Sequencing Panel
- ☐ Bardet Biedl Syndrome Deletion/Duplication Panel
- ☐ Joubert/Meckel Gruber Sequencing Panel
- ☐ Joubert/Meckel Gruber Deletion/Duplication Panel
- ☐ Meckel-Gruber Syndrome Sequencing Panel
- ☐ Meckel-Gruber Syndrome Deletion/Duplication Panel
- ☐ Nephronophthisis Sequencing Panel
- ☐ Nephronophthisis Deletion/Duplication Panel

Coffin Siris testing

- ☐ Coffin Siris Syndrome Sequencing Panel
- ☐ Coffin Siris Deletion/Duplication Panel

Congenital Muscle Disease testing

- ☐ Congenital Myopathy Sequencing Panel
- ☐ Congenital Myopathy Deletion/Duplication Panel
- ☐ Congenital Muscular Dystrophy Sequencing Panel
- ☐ Congenital Muscular Dystrophy Deletion/Duplication Panel
- ☐ Congenital Myasthenic Syndrome Sequencing Panel
- ☐ Congenital Myasthenic Syndrome Deletion/Duplication Panel
- ☐ Congenital Myopathy with Prominent Contractures Sequencing Panel
- ☐ Congenital Myopathy with Prominent Contractures Deletion/Duplication Panel
- ☐ Limb Girdle Muscular Dystrophy Sequencing Panel
- ☐ Limb Girdle Muscular Dystrophy Deletion/Duplication Panel
- ☐ Neuromuscular Disorders Sequencing Panel
- ☐ BIN1 Sequencing ☐ BIN1 Del/Dup
- ☐ DNM2 Sequencing ☐ DNM2 Del/Dup
- ☐ MTM1 Sequencing ☐ MTM1 Del/Dup
- ☐ RYR1 Sequencing ☐ RYR1 Del/Dup

Cornelia de Lange syndrome (CdLS) testing

- ☐ Cornelia de Lange Syndrome Panel
- ☐ Cornelia de Lange Syndrome PLUS Sequencing Panel
- ☐ NIPBL Sequencing ☐ NIPBL Del/Dup
- ☐ SMC1A Sequencing ☐ SMC1A Del/Dup
- ☐ Tier 3: SMC3, RAD21, HDAC8 sequencing
- ☐ Tier 3: SMC3, RAD21, HDAC8 deletion/duplication analysis

Craniofacial testing

- ☐ Craniofacial Sequencing Panel
- ☐ Craniofacial Deletion/Duplication Panel
- ☐ Facial Dysostosis Sequencing Panel
- ☐ Facial Dysostosis Deletion/Duplication Panel

TEST REQUESTS - Requisition Form

The University of Chicago Genetic Services Laboratories

Distal Arthrogryposes testing

- ☐ Distal Arthrogryposes Sequencing Panel
☐ Distal Arthrogryposes Deletion/Duplication Panel

Epilepsy testing

To order our Epilepsy Exome Panel, please use our Epilepsy Exome requisition form.

- ☐ Early Infantile Epileptic Encephalopathy Panel
☐ ARX Sequencing ☐ ARX Del/Dup
☐ STXBP1 Sequencing ☐ STXBP1 Del/Dup
☐ SLC25A22 Sequencing ☐ SLC25A22 Del/Dup
☐ SPTAN1 Sequencing ☐ SPTAN1 Del/Dup
☐ PCDH19 Sequencing ☐ PCDH19 Del/Dup

Hereditary Hemorrhagic Telangiectasia (HHT) testing

- ☐ Hereditary Hemorrhagic Telangiectasia (HHT) Sequencing Panel

Hyperinsulinism testing

Please see our endocrinology requisition form.

Intellectual disability (ID) testing

To order our Intellectual Disability Exome Panel, please use our Intellectual Disability Exome requisition form.

- ☐ Autosomal Recessive Non-Specific ID Sequencing Panel
☐ X-Linked Non-Specific ID Sequencing Panel
☐ Non-Specific ID Sequencing Panel

Kabuki syndrome

- ☐ Kabuki Syndrome Comprehensive Panel
☐ KMT2D (MLL2) Sequencing ☐ KMT2D (MLL2) Del/Dup
☐ KDM6A Sequencing ☐ KDM6A Del/Dup

Lipodystrophy testing

Please see our endocrinology requisition form.

Macrocephaly testing

- ☐ Macrocephaly Sequencing Panel
☐ Macrocephaly Deletion/Duplication Panel
☐ NSD1 Mutation Analysis
☐ NSD1 Sequencing ☐ NSD1 Del/Dup
☐ EZH2 Sequencing ☐ EZH2 Del/Dup
☐ NFIX Sequencing

MCT8 (Allan-Herndon-Dudley syndrome) testing

- ☐ Tier 1 (SLC16A2 (MCT8) Thyroid panel) followed by Tier 2 (SLC16A2 (MCT8) sequencing) if Tier 1 abnormal.

**3-10cc blood in an EDTA tube and 3-10cc blood in a red top tube required.

Microcephalic osteodysplastic primordial dwarfism

- ☐ Seckel Syndrome Sequencing Panel
☐ Seckel Syndrome Deletion/Duplication Panel
☐ Meier-Gorlin Syndrome Sequencing Panel
☐ Meier-Gorlin Syndrome Deletion/Duplication Panel
☐ Comprehensive Primordial Dwarfism Sequencing Panel
☐ Comprehensive Primordial Dwarfism Deletion/Duplication Panel
☐ PCNT Sequencing ☐ PCNT Del/Dup

Microcephaly testing

- ☐ Microcephaly Sequencing Panel
☐ Comprehensive Autosomal Recessive Primary Microcephaly Panel
☐ ASPM Sequencing ☐ ASPM Del/Dup
☐ IER3IP1 Sequencing ☐ IER3IP1 Del/Dup
☐ NDE1 Sequencing ☐ NDE1 Del/Dup
☐ PNKP Sequencing ☐ PNKP Del/Dup
☐ STAMBP Sequencing ☐ STAMBP Del/Dup
☐ WDR62 Sequencing ☐ WDR62 Del/Dup

Noonan syndrome

- ☐ Noonan Syndrome Sequencing Panel
☐ Noonan Syndrome Deletion/Duplication Panel

Neonatal Diabetes and Maturity-Onset Diabetes of the Young (MODY) testing

Please see our endocrinology requisition form.

Monogenic Obesity testing

- ☐ Monogenic Obesity Sequencing Panel

Multiple Congenital Anomalies testing

- ☐ Cytogenomic SNP array (postnatal)

Neurodegeneration with brain iron accumulation (NBIA) testing

- ☐ NBIA Sequencing Panel
☐ NBIA Deletion/Duplication Panel
☐ CP Sequencing ☐ CP Del/Dup
☐ FTL Sequencing ☐ FTL Del/Dup
☐ PANK2 Sequencing ☐ PANK2 Del/Dup
☐ PLA2G6 Sequencing ☐ PLA2G6 Del/Dup

Pancreatic Agenesis testing

Please see our endocrinology requisition form.

Prader-Willi syndrome testing

- ☐ Prader Willi Syndrome Series
☐ Methylation Specific-MLPA
☐ UPD15 (requires samples from both parents also)
☐ Imprinting Center Deletion Analysis
☐ MAGEL2 sequencing

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Rett/Atypical Rett syndrome testing

- ☐ Rett/Atypical Rett Syndrome Panel
- ☐ Rett/Angelman Syndrome Sequencing Panel
- ☐ Rett/Angelman Syndrome Deletion/Duplication Panel
- ☐ MECP2 Sequencing ☐ MECP2 Del/Dup
- ☐ CDKL5 Sequencing ☐ CDKL5 Del/Dup
- ☐ FOXP1 Sequencing ☐ FOXP1 Del/Dup
- ☐ MEF2C Sequencing ☐ MEF2C Del/Dup

Rubinstein-Taybi syndrome testing

- ☐ Rubinstein-Taybi Syndrome Series
- ☐ CREBBP Sequencing ☐ CREBBP Del/Dup
- ☐ EP300 Sequencing ☐ EP300 Del/Dup

UGT1A1 Testing

- ☐ UGT1A1 Genotyping for Gilbert syndrome
- ☐ UGT1A1 Genotyping for irinotecan dosing
- ☐ UGT1A1 Sequencing for Crigler-Najjar syndrome
- ☐ UGT1A1 Del/Dup (by array-CGH) for Crigler-Najjar syndrome

UPD Testing (Requires sample from both parents also)

- ☐ UPD6 ☐ UPD14
- ☐ UPD7 ☐ UPD15

Other Testing

Aceruloplasminemia

- ☐ CP Sequencing ☐ CP Del/Dup

Albinism

- ☐ Albinism Sequencing Panel
- ☐ Albinism Deletion/Duplication Panel

Alström syndrome

- ☐ ALMS1 Sequencing ☐ ALMS1 Del/Dup

Alternating Hemiplegia of Childhood

- ☐ ATP1A3 Sequencing

Aniridia

- ☐ PAX6 Sequencing ☐ PAX6 Del/Dup

Baraitser-Winter syndrome

- ☐ Baraitser Winter Syndrome Sequencing Panel
- ☐ Baraitser Winter Syndrome Deletion/Duplication Panel

Beckwith-Wiedemann syndrome/IMaGe syndrome

- ☐ CDKN1C Sequencing

Bernard-Soulier syndrome

- ☐ Gplb β Sequencing ☐ Gplb β Del/Dup

Charcot-Marie-Tooth disease

- ☐ DNM2 Sequencing ☐ DNM2 Del/Dup

CHARGE syndrome

- ☐ CHD7 Sequencing ☐ CHD7 Del/Dup

CHILD syndrome

- ☐ NSDHL Sequencing ☐ NSDHL Del/Dup

CHIME syndrome

- ☐ PIGL Sequencing ☐ PIGL Del/Dup

Hydroxyglutaric acidurias

- ☐ D-2 and L2-Hydroxyglutaric Aciduria Sequencing Panel
- ☐ D-2-Hydroxyglutaric Aciduria Sequencing Panel
- ☐ L2HGDH Sequencing
- ☐ SLC25A1 sequencing

Congenital heart defects (isolated)

- ☐ NKX2.5 Sequencing ☐ NKX2.5 Del/Dup

Congenital malabsorptive diarrhea

- ☐ NEUROG3 Sequencing ☐ NEUROG3 Del/Dup

Donnai-Barrow syndrome

- ☐ LRP2 Sequencing ☐ LRP2 Del/Dup

Exome Select

- ☐ Exome Select Custom Sequencing Panel (please contact us prior to ordering this test)

Fanconi-Bickel syndrome

- ☐ SLC2A2 Sequencing ☐ SLC2A2 Del/Dup

Floating Harbor syndrome

- ☐ SRCAP Sequencing ☐ SRCAP Del/Dup

Glucose transporter type 1 deficiency

- ☐ SLC2A1 Sequencing ☐ SLC2A1 Del/Dup

Goldberg Schprintzen megacolon syndrome

- ☐ KIAA1279 Sequencing ☐ KIAA1279 Del/Dup

Hearing loss

- ☐ GJB2 (CX26) Sequencing ☐ GJB2 (CX26) Del/Dup

Hereditary Breast and Ovarian Cancer

- ☐ Ashkenazi Jewish BRCA1/BRCA2 founder mutations

Hereditary mixed polyposis syndrome

- ☐ SCG5/GREM1 targeted duplication testing (founder mutation)

Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum

- ☐ SLC12A6 Sequencing ☐ SLC12A6 Del/Dup

Hypoinsulinemic Hypoglycemia with Hemihypertrophy

- ☐ AKT2 Sequencing ☐ AKT2 Del/Dup

Hyperinsulinism (Familial) testing

Please use our Hyperinsulinism specific requisition form to order testing.

IPEX syndrome (Immune dysregulation, polyendocrinopathy, enteropathy, X-linked)

- ☐ FOXP3 Sequencing ☐ FOXP3 Del/Dup

Laminopathies

- ☐ LMNA Sequencing ☐ LMNA Del/Dup

Marshall-Smith syndrome

- ☐ NFIX Sequencing

Menkes disease

- ☐ ATP7A Sequencing ☐ ATP7A Del/Dup

Mitchell-Riley syndrome

- ☐ RFX6 Sequencing ☐ RFX6 Del/Dup

Mowat-Wilson syndrome

- ☐ ZEB2 Sequencing ☐ ZEB2 Del/Dup

Neuronal Ceroid Lipofuscinoses (NCLs)

- ☐ Neuronal Ceroid Lipofuscinoses Panel

TEST REQUESTS - Requisition Form

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Nicolaides-Baraitser syndrome

- ☐ SMARCA2 Sequencing ☐ SMARCA2 Del/Dup

Oculodentodigital dysplasia (ODDD)

- ☐ GJA1 Sequencing

OFD1-related disorders

- ☐ OFD1 Sequencing ☐ OFD1 Del/Dup

Pigmented Hypertrichotic Dermatitis with Insulin-Dependent Diabetes Mellitus (PHID)

- ☐ SLC29A3 Sequencing ☐ SLC29A3 Del/Dup

Pitt-Hopkins syndrome

- ☐ TCF4 Sequencing ☐ TFC4 Del/Dup

Renal Cystic Disorders

- ☐ Renal Cystic Disorders Sequencing Panel

Roberts syndrome

- ☐ ESCO2 Sequencing ☐ ESCO2 Del/Dup

RNA testing

- ☐ Custom RNA Splicing Analysis (Please contact UCGS Lab Staff for prior approval before ordering. Requires fresh blood in PAX tube.)

Robinow syndrome

- ☐ ROR2 Sequencing ☐ ROR2 Del/Dup
☐ WNT5A Sequencing ☐ WNT5A Del/Dup

Schinzel-Giedion syndrome

- ☐ SETBP1 Sequencing ☐ SETBP1 Del/Dup

Temple-Baraitser syndrome

- ☐ KCNH1 Sequencing

SHORT syndrome

- ☐ PIK3R1 Sequencing

Thiamine Responsive Megaloblastic Anemia (TRMA)

- ☐ SLC19A2 Sequencing ☐ SLC19A2 Del/Dup

Type A Insulin Resistant Diabetes with Acanthosis Nigricans

- ☐ INSR Sequencing ☐ INSR Del/Dup

Warburg Micro syndrome

- ☐ Warburg Micro Syndrome Comprehensive Panel
☐ Warburg Micro Syndrome Sequencing Panel
☐ Warburg Micro Syndrome Deletion/Duplication Panel

Wiedemann-Steiner syndrome

- ☐ KMT2A (MLL) Sequencing ☐ KMT2A (MLL) Del/Dup

Wilson disease

- ☐ ATP7B Sequencing ☐ ATP7B Del/Dup

Wolcott-Rallison syndrome

- ☐ EIF2AK3 Sequencing ☐ EIF2AK3 Del/Dup

Wolfram syndrome

- ☐ Wolfram Syndrome Sequencing Panel
☐ Wolfram Syndrome Deletion/Duplication Panel

Woodhouse-Sakati syndrome

- ☐ DCAF17 Sequencing ☐ DCAF17 Del/Dup

Targeted Mutation Analysis

(Testing for a previously detected mutation or sequence change)

Requires prior approval by UCGS Lab Staff if this is a gene for which we do not offer full sequencing.

Gene: _____

Change: _____

Single Gene Sequence Analysis

Any gene included in one of our sequencing panels can also be ordered individually. Please contact UCGS Lab Staff for prior approval before ordering.

Gene Requested: _____

Single Gene Deletion/Duplication Analysis

Any gene included in one of our deletion/duplication panels can also be ordered individually. Please contact UCGS Lab Staff for prior approval before ordering.

Gene Requested: _____

BILLING OPTIONS

There are some tests for which we do not offer insurance billing. Please consult our website and quick guide (list of tests, costs, TAT and CPT codes) or contact us for more information.

All samples received with incomplete billing information will delay processing time.
Test cancelled while "in progress" will be billed for the amount of work completed up to that point.
Please forward all billing questions to: youtlaw@bsd.uchicago.edu or call (773-834-8220).

Patient Name: Last _____ First _____ (MI): _____ Date of Birth: _____

1.) Institutional Billing (Pre-payment is required for all samples referred from outside the US or Canada.)

Billing Institution: _____ PO#: _____

Financial Contact: _____ Phone: _____ Fax: _____

Address: _____ City: _____ State: _____ Zip: _____

Email (required): _____

2.) Self-Pay

We accept all major credit cards. Please call our office (773-834-8220) for credit card processing.



Important notice: We will not be responsible for refunding any "cost differential" that may occur as a result of a patient seeking any type of reimbursement.

☐ Wire Transfer (Please include 'Genetics Services Laboratories' and invoice numbers to ensure proper receipt.)

Electronic funding information, as follows: The Northern Trust Bank – (Physical Address) 50 S. LaSalle Street, Chicago, IL 60675
ABA/Routing No.: 071000152, International SWIFT Code: CNORUS44, University of Chicago Wire Account No.: 28509

Amount \$ _____ (USD) Date of Transfer: _____ Name of Institution: _____

☐ Check/Money Order (Make check/money order payable to: **The University of Chicago Genetic Services**) Amount Enclosed \$ _____
(Please note: All bank fees for returned checks will be added to the original charge of patient invoice)

3.) Insurance Billing (We do NOT accept Illinois or any out-of-state Medicaid. Please note we do not bill insurance for all our testing options. Please see our website for more details.) A legible photocopy of the front and back of the insurance card and insurance authorization must be included.

ICD-10 Diagnosis Code(s): _____ (Must be provided or insurance cannot be filed.)

Policyholder Name: _____ Date of Birth: ____/____/____ Gender: ☐ Male ☐ Female

Policyholder Address: _____ City: _____ State: _____ Zip: _____

Relationship to the Patient: ☐ Self ☐ Spouse ☐ Dependent ☐ Other Preauthorization # (if applicable): _____

Name of Primary Insurance: _____ Policy No. _____ Group No.: _____

Insurance Address: _____ City: _____ State: _____ Zip: _____

PCP/Referring Physician Name: _____ NPI #: _____

Name of Secondary Insurance: _____ Policy No.: _____ Group No.: _____

Insurance Address: _____ City: _____ State: _____ Zip: _____

The policy holder's signature to the following statement: I hereby authorize any physician who treated or attended to me or my dependent(s) to furnish any medical information requested. In consideration of services rendered, I hereby transfer and assign to the University of Chicago Genetic Services Laboratories any benefits of insurance I may have. I assume responsibility for the balance of the cost of testing not paid by my insurance company. A photocopy of this authorization shall be considered as effective and valid as original.

Authorized Signature: _____ Date: ____/____/____

RESEARCH CONSENT FORM – The University of Chicago

The Division of Biological Sciences | University of Chicago Medical Center

CONSENT/AUTHORIZATION BY SUBJECT FOR PARTICIPATION IN A RESEARCH PROTOCOL FOR THE BETTER UNDERSTANDING OF THEIR GENETIC CONDITION

Protocol Number: 11-0151

Name of Subject : _____

Date of Birth: _____

STUDY TITLE: Molecular Genetic Studies of Rare Orphan Genetic Disease

Research Team: Soma Das, Ph.D.
5841 S. Maryland Ave. Room L-155 MC 0077, Chicago, IL 60637
773-834-0555

You are being asked to allow your child to participate in a research study that may help us learn more about the genetic condition for which you are being tested. This consent form describes the study, the risks and benefits of participation, as well as how your confidentiality will be maintained. Please take your time to contact us with questions and feel comfortable making a decision whether to participate or not. If you decide to participate in this study, please sign this form. **Throughout this consent form, “you” will refer to you or your child, as appropriate.**

WHY IS THIS STUDY BEING DONE?

You have already consented to clinical genetic testing. We are asking you to also participate in further studies. The purpose of these studies is to learn more about the genetic cause of diseases tested for in our lab, gather more information about these disorders, and experiment with new methods that may be better for testing.

WHAT IS INVOLVED IN THE STUDY?

During this study, Dr. Das and her team will collect information about you for this research. We may contact your doctor to request additional Protected Health Information (PHI), which consists of any health information related to your diagnosis (such as date of birth, medical record number, primary diagnosis, clinical features, relevant and family history, outcome). The data collected will be used to develop a database of patients being tested for genetic diseases and will be kept for the duration of the database. This study will look at how often different genetic mutations happen and clinical information related to the mutation.

When our lab is researching new genes or testing methods that are related to your diagnosis, we may include your sample, with others from similar patients in a small study before offering this new test. This data will help in directing doctors about the likelihood of a positive or negative test result in their patient. We may also use your sample to set up new methods that will improve the clinical testing in our laboratory. Your clinical information and sample, without any

identifiers, may also be shared with other researchers that are interested in this specific condition.

HOW LONG WILL I BE IN THE STUDY?

Once enrolled, you will likely remain in this study as long as your DNA sample remains in our laboratory. If you want your sample, to be removed from the study at any time, please contact us, and the sample will not be used for further studies. Existing results will remain in our database until the study ends.

WHAT ARE THE RISKS OF THE STUDY?

There are no known added risks of the research. No additional information will be obtained from you, as all of the information has already been collected as part of clinical genetic testing or evaluation by your doctor.

ARE THERE ANY BENEFITS TO TAKING PART IN THE STUDY?

If you agree to take part in this study, there may be direct medical benefit to your family. We may identify a cause for the genetic disease in your family. If a mutation is identified in your DNA, through our testing, your referring doctor will be notified and will receive a clinical report. Our study may also be helpful in finding the genetic causes of disease and will benefit doctors and patients as a group.

WHAT OTHER OPTIONS ARE THERE?

You may choose not to participate.

WHAT ARE THE COSTS?

There will be no additional costs to you or your insurance company resulting from this research study. However, you or your insurance company will be responsible for costs related to your usual medical care.

WILL I BE PAID FOR MY PARTICIPATION?

You and your child will not be paid to participate.

WHAT ABOUT PRIVACY?

Study records that identify you will be kept private. All of your personal information will be entered into a password-protected database to prevent access to non-authorized personnel. If your data is shared with other researchers, all patient identifiers will be removed. Data from this study may be used in medical journals or presentations. If results from this study or related studies are made public in a medical journal, individual patients will not be identified. If we wish to use a patient's identity in a medical journal, we will ask for your permission at that time.

As part of the study, Dr. Das and her team will report any positive results of further testing to your referring doctor and/or genetic counselor. Dr. Das may also share these results, without your name or date of birth, with other researchers.

RESEARCH CONSENT FORM – The University of Chicago

The Division of Biological Sciences | University of Chicago Medical Center

People from the University of Chicago, including the Institutional Review Board (IRB), a committee that oversees research at the University of Chicago, may also view the records of the research. If health information is shared outside the University of Chicago, the same laws that the University of Chicago must obey may not protect your health information. Dr. Das does not have to give you any results that are not important to your health or your family's health at that time.

This consent form will be kept by the research team for at least six years. The study results will be kept in your child's research record and be used by the research team indefinitely. When the study ends, your personal information will be removed from all results. Any information shared with your doctor may be included in your medical record and kept forever.

WHAT ARE MY RIGHTS AS A PARTICIPANT?

Taking part in this study is optional. You may choose not to participate at any time during the study. Choosing not to participate or leaving the study will not affect your clinical testing at the University of Chicago.

If you choose to leave the study and you do not want any of your future health information to be used, you must inform Dr. Das in writing at the address on the first page. Dr. Das may still use your information that was collected before your written notice. You will be given a signed copy of this form. This consent form does not have an expiration date.

WHO DO I CALL IF I HAVE QUESTIONS OR PROBLEMS?

If you have further questions about the study, please call 773-834-0555.

If you have any questions about your rights in this research study you may contact the IRB, which protects participants in research projects. You may reach the Committee office between 8:30 am and 5:00 pm, Monday through Friday, by calling (773) 702-6505 or by writing: IRB, University of Chicago, 5751 S. Woodlawn Ave., McGiffert Hall, Chicago, Illinois 60637.

Consent

I have received information about this research project and the procedures. No guarantee has been given about possible results. I will receive a signed copy of this consent form for my records.

I give my permission to participate in the above research project.

Signature of Subject: _____

Date: _____

I give my permission for my child/relative/the person I represent to participate in the above research project.

Signature of Parent / Legal Guardian / Legally Authorized Representative:

Date: _____