



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: _____

Genetic Counselor: _____

Phone: _____ Fax: _____

Phone: _____ Fax: _____

Email: _____

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/UofC 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: 3-10cc blood in an EDTA (purple top) tube (unless otherwise indicated). DNA samples are only accepted if the DNA extraction or isolation was performed at a CLIA-certified laboratory.

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

ALL NEXT GENERATION SEQUENCING TESTS INCLUDE BOTH SEQUENCING AND DELETION/DUPLICATION ANALYSIS

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 Panel

Brain malformation testing

Cerebellar/Pontocerebellar Hypoplasia (PCH) testing

- Cerebellar/Pontocerebellar Hypoplasia Panel

Cerebral Cortical Malformation testing

- Cerebral Cortical Malformation Panel

Holoprosencephaly testing

- Holoprosencephaly Panel

Hydrocephalus testing

- Comprehensive Hydrocephalus Panel
- Autosomal Recessive Non-Syndromic Hydrocephalus Panel

Lissencephaly testing

- Lissencephaly Panel
- Cobblestone Lissencephaly Panel

Polymicrogyria testing

- Polymicrogyria Panel

Chondrodysplasia punctata testing

- ARSE Mutation Analysis
- EBP Mutation Analysis
- Rhizomelic Chondrodysplasia Punctata Panel

Ciliopathy Testing

- Bardet Biedl Syndrome Panel
- Joubert/Meckel Gruber Panel
- Meckel-Gruber Syndrome Panel
- Nephronophthisis Panel

Coffin Siris testing

- Coffin Siris Syndrome Panel

Congenital Muscle Disease testing

- Congenital Myopathy Panel
- Congenital Muscular Dystrophy Panel
- Congenital Myasthenic Syndrome Panel
- Congenital Myopathy with Prominent Contractures Panel
- Congenital Myopathy with Fiber-Type Disproportion Panel
- Congenital Muscular Dystrophy-Dystroglycanopathy Panel
- Centronuclear Myopathy Panel
- Emery-Dreifuss Muscular Dystrophy Panel
- Bethlem Myopathy and Ullrich Muscular Dystrophy Panel
- Limb Girdle Muscular Dystrophy Panel
- Multiminicore Disease Panel
- Myopathy with Tubular Aggregates Panel
- NemaLine Myopathy Panel
- Neuromuscular Myopathy Panel

Cornelia de Lange syndrome (CdLS) testing

- Cornelia de Lange Syndrome Mosaicism Panel (**Sequencing only)
- Cornelia de Lange Syndrome PLUS Panel

Craniofacial testing

- Craniofacial Panel
- Facial Dysostosis Panel

Currarino syndrome testing

- MNX1 Mutation Analysis

Distal Arthrogyroses testing

- Distal Arthrogyroses Panel

Epilepsy testing

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

- Early Infantile Epileptic Encephalopathy Panel

Hereditary Hemorrhagic Telangiectasia (HHT) testing

- Hereditary Hemorrhagic Telangiectasia (HHT) Panel

Hyperinsulinism testing

Please see our endocrinology requisition form.

Wilson disease

- ATP7B Mutation Analysis

Wolcott-Rallison syndrome

- EIF2AK3 Mutation Analysis

Wolfram syndrome

- Wolfram Syndrome Panel

Woodhouse-Sakati syndrome

- DCAF17 Mutation Analysis

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 Mutation Analysis (sequencing and deletion/duplication 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: _____

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Intellectual disability (ID) testing

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

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

Kabuki syndrome

- Kabuki Syndrome Panel

Lipodystrophy testing

Please see our endocrinology requisition form.

Macrocephaly testing

- Macrocephaly Panel

MCT8 (Allan-Herndon-Dudley syndrome) testing

Tier 1 (*SLC16A2* (MCT8) Thyroid panel) followed by Tier 2 (*SLC16A2* (MCT8) Mutation Analysis) 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 Panel
- Meier-Gorlin Syndrome Panel
- Primordial Dwarfism Panel

Microcephaly testing

- Microcephaly Panel
- Autosomal Recessive Primary Microcephaly Panel

Noonan syndrome

- Noonan Syndrome Panel

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

Please see our endocrinology requisition form.

Monogenic Obesity testing

- Monogenic Obesity Panel

Multiple Congenital Anomalies testing

- Cytogenomic SNP array (postnatal)

Neurodegeneration with brain iron accumulation (NBIA) testing

- NBIA Panel

Pancreatic Agenesis testing

Please see our endocrinology requisition form.

Prader-Willi syndrome testing

- Methylation Specific-MLPA
- UPD15 (requires samples from both parents also)
- Imprinting Center Deletion Analysis
- MAGEL2* Mutation Analysis

Rett/Atypical Rett syndrome testing

- Rett/Atypical Rett Syndrome Panel
- Rett/Angelman Syndrome Panel

Rubinstein-Taybi syndrome testing

- Rubinstein-Taybi Syndrome Panel

UGT1A1 Testing

- UGT1A1* Genotyping for Gilbert syndrome
- UGT1A1* Genotyping for irinotecan dosing
- UGT1A1* Mutation Analysis for Crigler-Najjar syndrome

UPD Testing (Requires sample from both parents also)

- UPD6 UPD14
- UPD7 UPD15

Other Testing

Aceruloplasminemia

- CP* Mutation Analysis

Albinism

- Albinism Panel

Alström syndrome

- ALMS1* Mutation Analysis

Aniridia

- PAX6* Mutation Analysis

Baraitser-Winter syndrome

- Baraitser Winter Syndrome Panel

Beckwith-Wiedemann syndrome/IMAge syndrome

- CDKN1C* Mutation Analysis

Bernard-Soulier syndrome

- Gplbβ* Mutation Analysis

CHARGE syndrome

- CHD7* Mutation Analysis

CHILD syndrome

- NSDHL* Mutation Analysis

CHIME syndrome

- PIGL* Mutation Analysis

Hydroxyglutaric acidurias

- D-2 and L2-Hydroxyglutaric Aciduria Panel
- D-2-Hydroxyglutaric Aciduria Panel

Donnai-Barrow syndrome

- LRP2* Mutation Analysis

Exome Select

- Exome Select (please contact us prior to ordering this test)

Fanconi-Bickel syndrome

- SLC2A2* Mutation Analysis

Floating Harbor syndrome

- SRCAP* Mutation Analysis

Glucose transporter type 1 deficiency

- SLC2A1* Mutation Analysis

Goldberg Schprintzen megacolon syndrome

- KIAA1279* Mutation Analysis

Hereditary Breast and Ovarian Cancer

- Ashkenazi Jewish *BRCA1/BRCA2* founder mutations

Hereditary mixed polyposis syndrome

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

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Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum

SLC12A6 Mutation Analysis

Hypoinsulinemic Hypoglycemia with Hemihypertrophy

AKT2 Mutation Analysis

Hyperinsulinism (Familial) testing

Please use our Hyperinsulinism specific requisition form to order testing.

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

FOXP3 Mutation Analysis

Laminopathies

LMNA Mutation Analysis

Marshall-Smith syndrome

NFIX Mutation Analysis

Menkes disease

ATP7A Mutation Analysis

Mitchell-Riley syndrome

RFX6 Mutation Analysis

Mowat-Wilson syndrome

ZEB2 Mutation Analysis

Neuronal Ceroid Lipofuscinoses (NCLs)

Neuronal Ceroid Lipofuscinoses Panel

Nicolaides-Baraitser syndrome

SMARCA2 Mutation Analysis

Oculodentodigital dysplasia (ODDD)

GJA1 Mutation Analysis

OFD1-related disorders

OFD1 Mutation Analysis

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

SLC29A3 Mutation Analysis

Pitt-Hopkins syndrome

TCF4 Mutation Analysis

Renal Cystic Disorders

Renal Cystic Disorders Panel

Roberts syndrome

ESCO2 Mutation Analysis

Robinow syndrome

Robinow Syndrome Panel

Schinzel-Giedion syndrome

SETBP1 Mutation Analysis

Temple-Baraitser syndrome

KCNH1 Mutation Analysis

SHORT syndrome

PIK3R1 Mutation Analysis

Thiamine Responsive Megaloblastic Anemia (TRMA)

SLC19A2 Mutation Analysis

Type A Insulin Resistant Diabetes with Acanthosis Nigricans

INSR Mutation Analysis

Warburg Micro syndrome

Warburg Micro Syndrome Panel

Weaver syndrome

EZH2 Mutation Analysis

Wiedemann-Steiner syndrome

KMT2A (MLL) Mutation Analysis



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. Prices listed on our website are not applicable for insurance billing, please contact us for insurance pricing information.) 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: ____/____/____

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.

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.

The Genetic Information Nondiscrimination Act (GINA) is a federal law that may help protect you from health insurance or employment discrimination based on genetic information. GINA is a federal law that will protect you in the following ways:

- Health insurance companies and group plans may not request genetic information from this research;
- Health insurance companies and group plans may not use your genetic information when making decisions regarding your eligibility or premiums;
- Employers with 15 or more employees may not use your genetic information when making a decision to hire, promote, or fire you or when setting the terms of your employment.

GINA does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination based on an already-diagnosed genetic condition or disease.

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 child's testing at the University of Chicago.

If you choose to leave the study and you do not want any of your child's 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 child's information that was collected before to 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: University of Chicago, Institutional Review Board, 5841 S. Maryland Ave., MC7132, I-625, Chicago, IL 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: _____