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:** _____ ICD9: _____

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: _____

Specimen Type: ☐ Peripheral Blood ☐ DNA ☐ Amniotic Fluid ☐ Chorionic Villi ☐ Product of Conception (POC)
☐ Culture: _____ ☐ Other: _____

Gestational Age: _____ **weeks by:** ☐ LMP ☐ Ultrasound

Specimen Requirements: **Routine Tests:** 3-10cc blood in an EDTA (purple top) tube (unless otherwise indicated).
Prenatal Tests: 20cc amniotic fluid, 20mgs chorionic villi or 2 T25 flasks of cultured cells.

For all prenatal testing, please send 3-10cc of mother's blood in an EDTA/Purple Top tube to rule out maternal cell contamination.

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/ICD9 study code *(required)*
☐ Completed Billing Information *(required)*
☐ Completed Research Consent Form *(recommended)*

For Office Use Only

The UNIVERSITY of CHICAGO
Genetic Services
www.dnatesting.uchicago.edu

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 (*MECP2, TCF4, SLC9A6, UBE3A*)
- ☐ Angelman Syndrome Series (MS-MLPA, Tier 2 Panel if negative)
- ☐ Rett/Angelman Syndrome Sequencing Panel (19 genes)
- ☐ Rett/Angelman Syndrome Deletion/Duplication Panel (19 genes)
- ☐ *UBE3A* Sequencing ☐ *UBE3A* Del/Dup
- ☐ *SLC9A6* Sequencing ☐ *SLC9A6* Del/Dup

Brain malformation testing

Cerebellar/Pontocerebellar Hypoplasia (PCH) testing

- ☐ Cerebellar/Pontocerebellar Hypoplasia Sequencing Panel (17 genes)
- ☐ Cerebellar/Pontocerebellar Hypoplasia Deletion/Duplication Panel (17 genes)
- ☐ *TSEN54* Sequencing ☐ *TSEN54* Del/Dup
- ☐ *CASK* Sequencing ☐ *CASK* Del/Dup
- ☐ *OPHN1* Sequencing ☐ *OPHN1* Del/Dup

Cerebral Cortical Malformation testing

- ☐ Cerebral Cortical Malformation Sequencing Panel (46 genes)
- ☐ Cerebral Cortical Malformation Deletion/Duplication Panel (25 genes)

Holoprosencephaly testing

- ☐ Holoprosencephaly Sequencing Panel (9 genes)
- ☐ Holoprosencephaly Deletion/Duplication Panel (9 genes)

Hydrocephalus testing

- ☐ *L1CAM* Sequencing ☐ *L1CAM* Del/Dup
- ☐ Autosomal Recessive Non-Syndromic Hydrocephalus Sequencing Panel (*CCDCC8C* and *MPDZ*)
- ☐ Autosomal Recessive Non-Syndromic Hydrocephalus Deletion/Duplication Panel (*CCDCC8C* and *MPDZ*)

Lissencephaly testing

- ☐ Comprehensive Lissencephaly Panel (26 genes seq and del/dup)
- ☐ Cobblestone Lissencephaly Sequencing Panel (18 genes)
- ☐ Cobblestone Lissencephaly Deletion/Duplication Panel (8 genes)
- ☐ *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 (15 genes)
- ☐ Polymicrogyria Deletion/Duplication Panel (15 genes)
- ☐ *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 Series (*PEX7* sequencing, then reflex to *GNPAT* and *AGPS* sequencing if negative)
- ☐ Rhizomelic Chondrodysplasia Punctata Sequencing Panel (3 genes)

Ciliopathy Testing

- ☐ Bardet Biedl Syndrome Sequencing Panel (16 genes)
- ☐ Bardet Biedl Syndrome Deletion/Duplication Panel (16 genes)
- ☐ Joubert/Meckel Gruber Sequencing Panel (26 genes)
- ☐ Joubert/Meckel Gruber Deletion/Duplication Panel (26 genes)
- ☐ Meckel-Gruber Syndrome Sequencing Panel (11 genes)
- ☐ Meckel-Gruber Syndrome Deletion/Duplication Panel (11 genes)
- ☐ Nephronophthisis Sequencing Panel (13 genes)
- ☐ Nephronophthisis Deletion/Duplication Panel (13 genes)

Coffin Siris testing

- ☐ Coffin Siris Syndrome Sequencing Panel (11 genes)
- ☐ Coffin Siris Deletion/Duplication Panel (6 genes)

Congenital Muscle Disease testing

- ☐ Congenital Myopathy Sequencing Panel (19 genes)
- ☐ Congenital Myopathy Deletion/Duplication Panel (17 genes)
- ☐ Congenital Muscular Dystrophy Sequencing Panel (24 genes)
- ☐ Congenital Muscular Dystrophy Deletion/Duplication Panel (23 genes)
- ☐ Congenital Myopathy with Prominent Contractures Sequencing Panel (11 genes)
- ☐ Congenital Myopathy with Prominent Contractures Deletion/Duplication Panel (11 genes)
- ☐ Limb Girdle Muscular Dystrophy Sequencing Panel (31 genes)
- ☐ Limb Girdle Muscular Dystrophy Deletion/Duplication Panel (25 genes)
- ☐ Congenital Myasthenic Syndrome Sequencing Panel (18 genes)
- ☐ Congenital Myasthenic Syndrome Deletion/Duplication Panel (13 genes)
- ☐ *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 PLUS Sequencing panel (21 genes seq)
- ☐ Cornelia de Lange Syndrome Series (*NIPBL* sequencing and deletion/duplication, reflex to *SMC1A* sequencing and deletion/duplication if negative, then reflex to Tier 3: *SMC3, RAD21, HDAC8* sequencing and deletion/duplication if negative)
- ☐ *NIPBL* Sequencing ☐ *NIPBL* Del/Dup
- ☐ *SMC1A* Sequencing ☐ *SMC1A* Del/Dup
- ☐ Tier 3:*SMC3, RAD21, HDAC8* sequencing
- ☐ Tier 3: *SMC3, RAD21, HDAC8* deletion/duplication analysis

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Craniofacial testing

- ☐ Craniofacial Sequencing Panel (21 genes)
- ☐ Craniofacial Deletion/Duplication Panel (21 genes)
- ☐ Facial Dysostosis Sequencing Panel (8 genes)
- ☐ Facial Dysostosis Deletion/Duplication Panel (17 genes)

Distal Arthrogryposes testing

- ☐ Distal Arthrogryposes Sequencing Panel (10 genes)
- ☐ Distal Arthrogryposes Deletion/Duplication Panel (9 genes)

Epilepsy testing

- ☐ Early Infantile Epileptic Encephalopathy Panel (30 genes seq & 21 genes del/dup)
- ☐ Infantile and Childhood Epilepsy Sequencing Panel (94 genes)
- ☐ *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

Hyperinsulinism testing

Please see our endocrinology requisition form.

Intellectual disability (ID) testing

- ☐ Autosomal Recessive Non-Specific ID Sequencing Panel (51 genes)
- ☐ X-Linked Non-Specific ID Sequencing Panel (77 genes)
- ☐ Comprehensive Non-Specific ID Sequencing Panel (170 genes)

Kabuki syndrome

- ☐ Kabuki Syndrome Series (*KMT2D (MLL2)* sequencing, reflex to *KMT2D* deletion/duplication if negative, reflex to *KDM6A* sequencing if negative, reflex to *KDM6A* deletion/duplication if negative)
- ☐ *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 (15 genes)
- ☐ Macrocephaly Deletion/Duplication Panel (15 genes)
- ☐ Sotos Syndrome Series (*NSD1* sequencing, then reflex to *NSD1* del/dup if negative)
- ☐ *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 (7 genes)
- ☐ Seckel Syndrome Deletion/Duplication Panel (7 genes)
- ☐ Meier-Gorlin Syndrome Sequencing Panel (5 genes)
- ☐ Meier-Gorlin Syndrome Deletion/Duplication Panel (5 genes)
- ☐ Comprehensive Primordial Dwarfism Sequencing Panel (16 genes)
- ☐ Comprehensive Primordial Dwarfism Deletion/Duplication Panel (14 genes)
- ☐ *PCNT* Sequencing ☐ *PCNT* Del/Dup

Microcephaly testing

- ☐ Microcephaly Sequencing Panel (56 genes)
- ☐ Autosomal Recessive Primary Microcephaly Series (*ASPM* sequencing and deletion/duplication, reflex to Autosomal Recessive Primary Microcephaly Tier 2 Sequencing and Deletion/Duplication panel if *ASPM* negative)
- ☐ *ASPM* Sequencing ☐ *ASPM* Del/Dup
- ☐ Autosomal Recessive Primary Microcephaly Tier 2 Sequencing Panel (18 genes)
- ☐ Autosomal Recessive Primary Microcephaly Tier 2 Deletion/Duplication Panel (16 genes)
- ☐ *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

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

Please see our endocrinology requisition form.

Monogenic Obesity testing

- ☐ Monogenic Obesity Sequencing Panel (29 genes)

Neurodegeneration with brain iron accumulation (NBIA) testing

- ☐ NBIA Sequencing Panel (9 genes)
- ☐ NBIA Deletion/Duplication Panel (9 genes)
- ☐ *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, reflex to MAGEL2 sequencing if negative)
- ☐ 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 (*MECP2*, *CDKL5*, *MEF2C* , *FOXG1*)
- ☐ Rett/Angelman Syndrome Sequencing Panel (21 genes)
- ☐ Rett/Angelman Syndrome Deletion/Duplication Panel (19 genes)
- ☐ *MECP2* Sequencing ☐ *MECP2* Del/Dup
- ☐ *CDKL5* Sequencing ☐ *CDKL5* Del/Dup
- ☐ *FOXG1* Sequencing ☐ *FOXG1* Del/Dup
- ☐ *MEF2C* Sequencing ☐ *MEF2C* Del/Dup

Rubinstein-Taybi syndrome testing

- ☐ Rubinstein-Taybi Syndrome Series (*CREBBP* del/dup, then reflex to *CREBBP* sequencing if negative, then *EP300* sequencing if negative, then *EP300* del/dup if negative)
- ☐ *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 (20 genes)
- ☐ Albinism Deletion/Duplication Panel (20 genes)

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 (*ACTB* and *ACTG1*)
- ☐ Baraitser Winter Syndrome Deletion/Duplication Panel (*ACTB* and *ACTG1*)

Beckwith-Wiedemann syndrome/IMAGe syndrome

- ☐ *CDKN1C* Sequencing

Bernard-Soulier syndrome

- ☐ *GpIbβ* Sequencing ☐ *GpIbβ* 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

Combined D-2 and L-2-hydroxyglutaric aciduria

- ☐ *SLC25A1* sequencing

Congenital heart defects (isolated)

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

Congenital malabsorptive diarrhea

- ☐ *NEUROG3* Sequencing ☐ *NEUROG3* Del/Dup

D-2-hydroxyglutaric aciduria

- ☐ D-2-Hydroxyglutaric Aciduria Sequencing Panel *(D2HGDH* and *IDH2)*

Donnai-Barrow syndrome

- ☐ *LRP2* Sequencing ☐ *LRP2* Del/Dup

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

L-2-hydroxyglutaric aciduria

- ☐ *L2HGDH* Sequencing

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 (8 genes seq and del/dup)

Nicolaides-Baraitser syndrome
☐ *SMARCA2* Sequencing ☐ *SMARCA2* Del/Dup

Oculodentodigital dysplasia (ODDD)
☐ *GJA1* Sequencing

OFD1-related disorders
☐ *OFD1* Sequencing ☐ *OFD1* Del/Dup

Pigmented Hypertrichotic Dermatosis with Insulin-Dependent Diabetes Mellitus (PHID)
☐ *SLC29A3* Sequencing ☐ *SLC29A3* Del/Dup

Pitt-Hopkins syndrome
☐ *TCF4* Sequencing ☐ *TFC4* Del/Dup

Roberts syndrome
☐ *ESCO2* Sequencing ☐ *ESCO2* Del/Dup

Robinow syndrome
☐ *ROR2* Sequencing ☐ *ROR2* Del/Dup
☐ *WNT5A* Sequencing ☐ *WNT5A* Del/Dup

Schinzel-Giedion syndrome
☐ *SETBP1* Sequencing ☐ *SETBP1* Del/Dup

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 Sequencing Panel (*RAB18, RAB3GAP1, RAB3GAP2, TBC1D20*)
☐ Warburg Micro Syndrome Deletion/Duplication Panel (*RAB18, RAB3GAP1, RAB3GAP2, TBC1D20*)

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 (*CISD2* and *WFS1*)
☐ Wolfram Syndrome Deletion/Duplication Panel (*CISD2* and *WFS1*)

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:_____

The UNIVERSITY of CHICAGO
Genetic Services
www.dnatesting.uchicago.edu

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: _____

E-mail Invoice(s): ☐ Yes ☐ No 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-9 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 your child is 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.

Why is this study being done?

You have already consented to clinical genetic testing for your child. 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 your child for this research. We may contact your doctor for additional Protected Health Information (PHI) about your child, which consists of any health information related to your child's diagnosis (such as date of birth, medical record number, primary diagnosis, clinical features, relevant and family history, and 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 child's diagnosis, we may include your child's 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 child's sample to set up new methods that will improve the clinical testing in our laboratory. Your child's 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?

Your child will likely remain in this study as long as his/her DNA sample remains in our laboratory. If you want your child's 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 your child 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 child's DNA through our testing, your child's 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 your child 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 your child will be kept private. All of your child's personal information will be entered into a password-protected database to prevent access to non-authorized personnel. If your child's 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 child's 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 child's health information. Dr. Das does not have to give you any results that are not are not important to your child's or your family's health at that time.

The research team will keep this consent form 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 child's personal information will be removed from all results. Any information shared with your child's doctor may be included in your child's 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 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: IRB, University of Chicago, 5751 S. Woodlawn Ave., McGiffert Hall, Chicago, Illinois 60637.

Consent

PARENT/GUARDIAN/ OR LEGALLY AUTHORIZED REPRESENTATIVE:

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

Signature of Parent/Guardian/ Legally Authorized Representative:

_____

Date: _____

WITNESS

Signature of Witness:

_____

Date: _____