

PATIENT INFORMATION / DEMOGRAPHICS

PATIENT D.O.B. \_\_\_\_\_

PATIENT LAST NAME \_\_\_\_\_ FIRST NAME \_\_\_\_\_ MI \_\_\_\_\_

STREET ADDRESS \_\_\_\_\_

CITY \_\_\_\_\_ STATE \_\_\_\_\_ ZIP \_\_\_\_\_

SEX: M  F  \_\_\_\_\_

DUKE HISTORY # \_\_\_\_\_

PATIENT TELEPHONE #: \_\_\_\_\_

REQUESTING INSTITUTION PATIENT ID CODE: \_\_\_\_\_

PLEASE PROVIDE NAMES OF OTHER FAMILY MEMBERS BEING TESTED OR TESTED POSITIVE PREVIOUSLY

NAME \_\_\_\_\_ RELATIONSHIP \_\_\_\_\_

RESULTS (Including known mutations): \_\_\_\_\_

NAME \_\_\_\_\_ RELATIONSHIP \_\_\_\_\_

RESULTS (Including known mutations): \_\_\_\_\_

NAME \_\_\_\_\_ RELATIONSHIP \_\_\_\_\_

RESULTS (Including known mutations): \_\_\_\_\_

CLINICAL INFORMATION

ICD-9 CODE (Required): \_\_\_\_\_

INDICATION FOR TESTING / CLINICAL FINDINGS (Required): \_\_\_\_\_

ENZYME TESTING:  CONCURRENT  PREVIOUS (if applicable)

PERFORMING LAB: \_\_\_\_\_

ENZYME: \_\_\_\_\_

RESULTS: \_\_\_\_\_

SPECIMEN COLLECTION

SPECIMEN REQUIREMENTS:  
 PERIPHERAL BLOOD: 5cc (1-2cc for infants) in EDTA (purple-top tube)  
 CULTURED CELLS: T25 flask (80-90% confluent)  
**Please call the laboratory for requirements for other specimen types.**  
**\*\*Prenatal specimens must be accompanied by a maternal blood sample to rule out maternal cell contamination. Please call the laboratory for all prenatal samples**

COLLECTION DATE: \_\_\_\_/\_\_\_\_/\_\_\_\_

SPECIMEN TYPE:

Peripheral Blood

Skin biopsy (culturing fees apply)

Cultured fibroblasts

Amniotic fluid (culturing fees apply)

Cultured amniocytes / cultured chorionic villi

Extracted DNA\*\*\*\* Concentration: \_\_\_\_\_

OTHER (specify): \_\_\_\_\_

\*\*\*\*If submitting extracted DNA, please include CLIA number of lab that performed the extraction. DNA cannot be accepted from laboratories that are not CLIA-accredited.

BILLING / REPORTING INFORMATION

PHYSICIAN NAME AND ADDRESS: (Results will be sent to this address) \_\_\_\_\_

REQUESTING PHYSICIAN (PLEASE PRINT) \_\_\_\_\_

REQUESTING PHYSICIAN SIGNATURE (REQUIRED) \_\_\_\_\_

STREET ADDRESS \_\_\_\_\_

CITY \_\_\_\_\_ STATE \_\_\_\_\_ ZIP \_\_\_\_\_

TELEPHONE # \_\_\_\_\_ FAX # \_\_\_\_\_

EMAIL \_\_\_\_\_

**The DUHS Molecular Diagnostics Laboratory does not bill patients or their insurance companies. The submitting physician/institution is charged for all non-Duke patient testing.**

**BILLING CONTACT NAME AND ADDRESS: (Required)**

INSTITUTION / CONTACT PERSON \_\_\_\_\_

STREET ADDRESS \_\_\_\_\_

CITY \_\_\_\_\_ STATE \_\_\_\_\_ ZIP \_\_\_\_\_

TELEPHONE # \_\_\_\_\_ FAX # \_\_\_\_\_

TESTS REQUESTED

**Please note that informed consent is required for all genetic testing (to be obtained by the ordering physician)**

SLC37A4 SEQUENCING (Glycogen storage disease Ib)

GAA SEQUENCING (Pompe, Glycogen storage disease II)

AGL SEQUENCING (Glycogen storage disease III)

PHKA2 SEQUENCING (Glycogen storage disease IXa, XLG)

PHKG2 SEQUENCING (Glycogen storage disease IXc)

GLA SEQUENCING (Fabry disease)

TMRSS6 SEQUENCING (IRIDA)

TP53 SEQUENCING (Li-Fraumeni Syndrome)

MYH9 SEQUENCING

MOCS1 SEQUENCING (Molybdenum cofactor deficiency)

MOCS2 SEQUENCING (Molybdenum cofactor deficiency)

SUOX SEQUENCING (Isolated sulfite oxidase deficiency)

ALPL SEQUENCING (Hypophosphatasia (HPP))

IDUA SEQUENCING (Mucopolysaccharidosis I (MPS-1))

ABCD1 SEQUENCING (X-linked Adrenoleukodystrophy (X-ALD))

\*For the above tests please indicate one of the following:

FULL GENE (all coding exons)

ONE EXON: \_\_\_\_\_

TWO EXONS: \_\_\_\_\_ and \_\_\_\_\_

SHIPPING INFORMATION

Samples being concurrently tested for enzyme activity by the Duke Biochemical Genetics Laboratory will be shared between the two laboratories. Samples should be shipped via overnight courier to:

Duke Pediatric Biochemical Genetics Laboratory  
 801 Capitola Drive, Suite 6  
 Durham, NC 27713  
 919-549-0445 (phone) / 919-549-0709 (fax)

**Samples for genetic sequencing only should be sent to the address on the top of this form.**