

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): _____

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)

G6PC SEQUENCING (Glycogen storage disease Ia)

SLC37A4 SEQUENCING (Glycogen storage disease Ib)

GAA SEQUENCING (Pompe, Glycogen storage disease II)

AGL SEQUENCING (Glycogen storage disease III)

GBE1 SEQUENCING (Glycogen storage disease IV)

PHKA2 SEQUENCING (Glycogen storage disease IXa, XLG)

PHKG2 SEQUENCING (Glycogen storage disease IXc)

GLA SEQUENCING (Fabry disease)

Tmprss6 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))

Glycogen Storage Disease Gene Panel (25 genes) – for additional information, please visit www.dukemolecular.org

*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.