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**LABORATORY OF GENETICS AND GENOMICS**
For local courier service and/or inquiries, please contact 513-636-4474 • Fax: 513-636-4373
www.cincinnatichildrens.org/moleculargenetics • Email: LabGeneticCounselors@cchmc.org

**Mailing Address:**
3333 Burnet Avenue, Room R1042
Cincinnati, OH 45229

# LYSOSOMAL STORAGE DISEASE TEST REQUISITION

**All Information Must Be Completed Before Sample Can Be Processed**

## PATIENT INFORMATION

Patient Name: _____ , _____ , _____
Last First MI

Address: _____

_____

Home Phone: _____

MR# _____ Date of Birth _____ / _____ / _____

Gender: ☐ Male ☐ Female

## ETHNIC/RACIAL BACKGROUND (Choose All)

☐ European American (White)
☐ African-American (Black)
☐ Native American or Alaskan
☐ Asian-American
☐ Pacific Islander
☐ Ashkenazi Jewish ancestry
☐ Latino-Hispanic _____
(specify country/region of origin)
☐ Other _____
(specify country/region of origin)

## BILLING INFORMATION (Choose ONE method of payment)

### ☐ REFERRING INSTITUTION

Institution: _____

Address: _____

City/State/Zip: _____

Accounts Payable Contact Name: _____

Phone: _____

Fax: _____

Email: _____

### ☐ COMMERCIAL INSURANCE*

**Insurance can only be billed if requested at the time of service.**

Policy Holder Name: _____

Gender: _____ Date of Birth _____ / _____ / _____

Authorization Number: _____

Insurance ID Number: _____

Insurance Name: _____

Insurance Address: _____

City/State/Zip: _____

Insurance Phone Number: _____

*** PLEASE NOTE:**

- We will not bill Medicaid, Medicaid HMO, or Medicare except for the following: Cincinnati Children's Patients, Cincinnati Children's Providers, or Designated Regional Counties.
- If you have questions, please call 1-866-450-4198 for complete details.

## SAMPLE/SPECIMEN INFORMATION

SPECIMEN TYPE: ☐ Amniotic fluid ☐ Blood ☐ Cytobrushes ☐ Saliva
☐ Cord blood ☐ CVS ☐ Bone marrow ☐ Other: _____
☐ Tissue (specify): _____

Specimen Date: _____ / _____ / _____ Time: _____

Specimen Amount: _____

Each test requires 3 mL of whole blood in EDTA tube. Please call before sending alternate tissue samples, and for free cytobrush or saliva collection kits.

DRAWN BY: _____
*Phlebotomist must initial tube of specimen to confirm sample identity

## REFERRING PHYSICIAN

Physician Name (print): _____

Address: _____

Phone: ( _____ ) _____ Fax: ( _____ ) _____

Email: _____

Genetic Counselor/Lab Contact Name: _____

Phone: ( _____ ) _____ Fax: ( _____ ) _____

Email: _____

_____ Date: ____/ _____/ ____
**Referring Physician Signature (REQUIRED)**

☐ Patient signed completed ABN

Medical Necessity Regulations: At the government's request, the Molecular Genetics Laboratories would like to remind all physicians that when ordering tests that will be paid under federal health care programs, including Medicare and Medicaid programs, that these programs will pay only for those tests the relevant program deems to be (1) included as covered services, (2) reasonable, (3) medically necessary for the treatment and diagnosis of the patient, and (4) not for screening purposes.

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Patient Name: _____ Date of Birth: _____

## INDICATIONS/DIAGNOSIS/ICD-9 CODE

☐ Mutation detection in suspected affected patient

☐ Carrier (Heterozygote) testing

☐ Prenatal diagnosis (by previous arrangement only)

## PEDIGREE OR FAMILY HISTORY

Parental Consanguinity ☐ Y ☐ N

## TEST(S) REQUESTED

### Cystinosis

☐ *CTNS* full gene sequencing only

☐ *CTNS* 57 kb deletion analysis

☐ *CTNS* 57 kb deletion analysis with reflex to full gene sequencing

### Fabry Disease

☐ *GLA* full gene sequencing

☐ Reflex to deletion/duplication of *GLA*

### Gaucher Disease

☐ *GBA* full gene sequencing

### MPS II - Hunter syndrome

☐ *IDS* full gene sequencing

☐ Reflex to deletion/duplication of *IDS*

### Pompe Disease

☐ *GAA* full gene sequencing for Infantile Pompe disease (by prior arrangement only)

☐ *GAA* full gene sequencing

☐ Reflex to deletion/duplication of *GAA*

☐ **Targeted (family specific) mutation analysis of genes listed above**

Gene of interest _____

Proband's name _____

Proband's DOB _____

Proband's mutation _____

Relationship to proband _____

**Please call 513-636-4474 to discuss any family-specific mutation analysis with genetic counselor prior to shipment.**

**If testing was not performed at Cincinnati Children's, please include proband's report and at least 100ng of proband's DNA to use as a positive control.**

## CUSTOM GENE SEQUENCING

**Gene(s) to be sequenced (specify):** _____

_____

_____

**Only genes with clear published functional relationship to rare diseases are accepted.**

**Suspected syndrome/ condition:** _____

**Please choose one of the following:**

☐ Full gene(s) sequencing

☐ Full gene(s) sequencing with reflex to deletion and duplication analysis, if indicated **(please see list of genes available for del/dup at www.cincinnatichildrens.org/deldup)**

☐ Familial mutation analysis

Proband's name: _____

Proband's DOB: _____

Proband's mutation: _____

Patient's relation to proband: _____

**If testing was not performed at Cincinnati Children's, please include proband's report and at least 100ng of proband's DNA to use as a positive control.**

## DELETION AND DUPLICATION ASSAY

**Gene(s) to be analyzed (specify):** _____

_____

**Please see list of available genes at: www.cincinnatichildrens.org/deldup**

**Suspected syndrome/ condition:** _____

**Please choose one of the following:**

☐ Deletion and duplication analysis of gene(s) specified above

☐ Deletion and duplication analysis of gene(s) specified above with reflex to sequencing, if indicated

☐ Analysis of gene(s) specified above from previously analyzed deletion and duplication

☐ Familial deletion analysis

Proband's name: _____

Proband's DOB: _____

Proband's mutation: _____

Patient's relation to proband: _____

**If testing was not performed at Cincinnati Children's, please include proband's report and at least 100ng of proband's DNA to use as a positive control.**

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Patient Name: _____ Date of Birth: _____

## PRESENTING SYMPTOMS/HISTORY

### Cystinosis

Age at onset of symptoms: _____

- ☐ Growth retardation
- ☐ Renal failure
- ☐ Renal tubular fanconi syndrome
- ☐ Corneal cystine crystals
- ☐ Hypothyroidism
- ☐ Diabetes mellitus
- ☐ Hypogonadism
- ☐ Abnormal cystine concentration in leukocytes
- ☐ Other (please specify): _____

### Fabry Disease

Age at onset of symptoms: _____

- ☐ Acroparasthesias
- ☐ Angiokeratomas
- ☐ Cardiac disease
- ☐ Temperature intolerance
- ☐ History of stroke
- ☐ Renal disease
- ☐ Nausea, diarrhea, abdominal pain
- ☐ Pain in the _____
- ☐ Confirmed deficiency of α-galactosidase
- ☐ Other (please specify): _____

### Gaucher Disease

Age at onset of symptoms: _____

- ☐ Hepatosplenomegaly
- ☐ Bone manifestations
- ☐ Anemia
- ☐ Thrombocytopenia
- ☐ CNS involvement
- ☐ Confirmed deficiency of acid α-glucosidase
- ☐ Other (please specify): _____

### MPS II — Hunter syndrome

Age at onset of symptoms: _____

- ☐ Coarse facial features
- ☐ Hepatosplenomegaly
- ☐ Abdominal or inguinal hernias
- ☐ Developmental delay, hyperactivity, irritability
- ☐ Skeletal abnormalities
- ☐ Frequent ear & sinus infections, rhinorrhea, noisy breathing
- ☐ Increased dermatan sulfate and heparan sulfate in urine
- ☐ Confirmed deficiency of iduronate-2-sulfatase
- ☐ Other (please specify): _____

### Pompe Disease

Age at onset of symptoms: _____

- ☐ Hypertrophic cardiomyopathy
- ☐ Feeding difficulties
- ☐ Cardiac disease/failure
- ☐ Respiratory distress
- ☐ Muscle weakness
- ☐ Enlarged tongue
- ☐ Confirmed deficiency of acid α-glucosidase
- ☐ Other (please specify): _____