Cincinnati Children's

MOLECULAR GENETICS LABORATORY

For courier service and/or inquiries, please contact 513-636-4474 • Fax: 513-636-4373
www.cincinnatichildrens.org/moleculargenetics • Email: moleculargenetics@cchmc.org

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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: CCHMC Patients, CCHMC 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: _____

INDICATION FOR TESTING

☐ 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

☐ Ashkenazi Panel **(TESTS ONLY: N370S, L444P, 84GG, IVS2+1G>A)**

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.

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)**

☐ Targeted analysis for a common mutation seen in the population of interest

Mutation: _____

☐ Familial mutation analysis

Proband's name: _____

Proband's DOB: _____

Proband's mutation: _____

Patient's relation to proband: _____

Please include proband's report, if not performed at CCHMC.

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

☐ Targeted deletion or duplication analysis for a common mutation seen in the population of interest

Mutation: _____

☐ 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: _____

Please include proband's report, if not performed at CCHMC.

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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):_____