Cincinnati Children's
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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

INBORN ERRORS OF METABOLISM 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
☐ Cord blood ☐ CVS ☐ Bone marrow ☐ Saliva
☐ 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)

Contact Information for Results/Questions (*if different than ordering provider*):

Name & Title: _____

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

Email: _____

☐ **Patient signed completed ABN**

Medical Necessity Regulations: At the government's request, the Laboratory of Genetics and Genomics 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

Diagnosis/ICD-9 CODE: _____

Reason for Testing:

- ☐ Diagnosis in symptomatic patient
- ☐ Asymptomatic infant with abnormal newborn screen
- ☐ Carrier testing
- ☐ Presymptomatic diagnosis of at-risk sibling
- ☐ Prenatal testing (by previous arrangement only)
- ☐ Family histor of disease

Please specify relationship (e.g.; cousin): _____

PEDIGREE OR FAMILY HISTORY

Parental Consanguinity ☐ Y ☐ N

TEST(S) REQUESTED

☐ **MetaboSeq® Next Generation Sequencing Panel**
(sequencing of 56 genes including *ACAD9, ACADM, ACADS, ACADVL, ACAT1, AGL, ALDOA, ALDOB, CPT1A, CPT2, DECR1, ENO3, ETFA, ETFB, ETFDH, FBP1, G6PC, GAA, GBE1, GLUD1, GYS1, GYS2, HADH, HADHA, HADHB, HMGCL, HSD17B10, LAMP2, LPIN1, MLYCD, MPI, NADK2, OXCT1, PC, PCK1, PCK2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PPARG, PRKAG2, PYGL, PYGM, SLC22A5, SLC25A20, SLC2A2, SLC37A4, SLC52A2, SLC52A3, TANGO2, TAZ*)

- ☐ Reflex to deletion/duplication of *ACAD9, ACADM, ACADS, ACADVL, ACAT1, AGL, ALDOA, ALDOB, CPT1A, CPT2, DECR1, ENO3, ETFA, ETFB, ETFDH, FBP1, G6PC, GAA, GBE1, GYS1, GYS2, HADH, HADHA, HADHB, HMGCL, HSD17B10, LAMP2, LPIN1, MLYCD, MPI, OXCT1, PC, PCK1, PCK2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PPARG, PRKAG2, PYGL, PYGM, SLC22A5, SLC25A20, SLC2A2, SLC52A2, SLC52A3, TANGO2, TAZ)*
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____

_____

☐ **Glycogen Storage Disease Gene Sequencing Panel**
(sequencing of 19 genes including *AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYS1, GYS2, PFKM, PGAM2, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4*)

- ☐ Reflex to Metaboseq panel if results are non-diagnostic
- ☐ Reflex to deletion/duplication of *AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYS1, GYS2, PFKM, PGAM2, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM,* and *SLC2A2*
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____

_____

☐ **Riboflavin Disorders Gene Sequencing Panel**
(sequencing of 5 genes including *ETFA, ETFB, ETFDH, SLC52A2, SLC52A3*)

- ☐ Reflex to Metaboseq panel if results are non-diagnostic
- ☐ Reflex to deletion/duplication of entire panel
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____

_____

☐ **Elevated C16 Gene Sequencing Panel**
(sequencing of *SLC25A20* and *CPT2*)

- ☐ Reflex to deletion/duplication of *SLC25A20* and *CPT2*

☐ **LCHAD/TFP Gene Sequencing Panel for Long Chain 3 Hydroxyacyl CoA Dehydrogenase (LCHAD) Deficiency / Trifunctional Protein Deficiency (TFP)**
(sequencing of *HADHA* and *HADHB*)

- ☐ Reflex to deletion/duplication of *HADHA* and *HADHB*

☐ **GSD type I Gene Sequencing Panel**
(sequencing of *G6PC* and *SLC37A4*)

- ☐ Reflex to deletion/duplication of *G6PC*†

Single Gene Testing
Note: Single gene sequencing is available for all genes in the MetaboSeq Panel. Please select a gene from the list below, or use the Custom Gene Sequencing section for any gene that is not specified below.

Medium Chain ACYL-COA Dehydrogenase (MCAD) Deficiency

- ☐ *ACADM* (K329E) genotyping only
- ☐ *ACADM* (K329E) genotyping, with reflex to full *ACADM* sequencing, if indicated
- ☐ *ACADM* full gene sequence analysis
  - ☐ Reflex to *ACADM* deletion/duplication analysis

Very Long Chain ACYL-COA Dehydrogenase (VLCAD) Deficiency

- ☐ *ACADVL* full gene sequence analysis
  - ☐ Reflex to *ACADVL* deletion/duplication analysis

Hereditary Fructose Intolerance

- ☐ *ALDOB* full gene sequence analysis
  - ☐ Reflex to *ALDOB* deletion/duplication analysis

Carnitine Palmitoyltransferase 2 (CPT2) Deficiency

- ☐ *CPT2* full gene sequencing analysis
  - ☐ Reflex to *CPT2* deletion/duplication analysis

Carnitine Deficiency, Systemic Primary (CDSP)

- ☐ *SLC22A5* full gene sequencing analysis
  - ☐ Reflex to *SLC22A5* deletion/duplication analysis

Pompe Disease

- ☐ *GAA* full gene sequence analysis
  - ☐ Reflex to *GAA* deletion/duplication analysis

- ☐ Targeted (family specific) variant analysis of genes listed above
  - Gene of interest _____
  - Proband's name _____
  - Proband's DOB _____
  - Proband's variant _____
  - Relationship to proband _____

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

If testing was <u>not</u> 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/Duplication analysis of GLUD1, NADK2 and SLC37A4 is not available at this time.

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Patient Name: _____ Date of Birth: _____

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 variant analysis

Proband's name: _____

Proband's DOB: _____

Proband's variant: _____

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/duplication analysis

Proband's name: _____

Proband's DOB: _____

Proband's deletion/duplication: _____

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: _____

Clinical History is Required

CLINICAL HISTORY

General

☐ Lethargy
☐ Vomiting
☐ Failure to thrive
☐ Respiratory insufficiency/failure
☐ Sudden unexplained infant death
☐ Coma

Metabolic

☐ Abnormal acylcarnitine profile (specify results) _____

_____

☐ Abnormal newborn screen (list disease suggested) _____

_____

☐ Hypoketotic hypoglycemia
☐ Hyperinsulinemic hypoglycemia
☐ Hypoglycemia
☐ Diabetes
☐ Lipodystrophy
☐ Low ketone body formation
☐ Hyperammonemia
☐ Elevated serum creatine kinase
☐ Metabolic acidosis

Neuro/Muscular

☐ Hypotonia
☐ Brain dysplasia
☐ Skeletal/facial myopathy
☐ Exercise-induced myopathy
☐ Rhabdomyolysis
☐ Neuropathy
☐ Seizures
☐ Choreoathetosis
☐ Developmental delay
☐ Progressive peripheral neuropathy
☐ Myalgias

Cardiovascular

☐ Arrhythmia (specify type) _____

_____

☐ Hypertension
☐ Cardiomyopathy
☐ Cardiomegaly
☐ Coagulopathy
☐ Cardiac failure
☐ Pericardial effusion

Eye

☐ Retinopathy

Liver

☐ Elevated liver enzymes
☐ Liver dysfunction/failure
☐ Hepatic encephalopathy
☐ Hepatomegaly/enlarged liver
☐ Reye syndrome-like phenotype

Maternal complications during pregnancy

☐ Preeclampsia
☐ Hyperemesis gravidarum
☐ Acute fatty liver of pregnancy
☐ HELLP syndrome

Congenital abnormalities/malformations /dysmorphic features

(Please specify)

_____
_____
_____
_____

Other Symptoms *(Please specify)*

_____
_____
_____
_____