

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)

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.

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 history of disease

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

PEDIGREE OR FAMILY HISTORY

Parental Consanguinity Y N

TEST(S) REQUESTED

Panels

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, ETFA, ETFB, ETFDH, FBP1, G6PC, GAA, GBE1, GYS2, HADH, HADHA, HADHB, HMGCL, HSD17B10, LAMP2, MLYCD, MPI, PC, PFKM, PGK1, PHKA2, PHKB, PHKG2, PPARG, PYGM, SLC22A5, SLC25A20, SLC37A4, and 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, G6PC, GAA, GBE1, GYS2, PFKM, PHKA2, PHKB, PHKG2, PYGM, and SLC37A4*

Reflex to deletion/duplication of single gene(s) (specify): _____

Riboflavin Disorders Gene Sequencing Panel

(sequencing of 5 genes including *ETF A, ETFB, ETFDH, SLC52A2, SLC52A3*)

Reflex to Metaboseq panel if results are non-diagnostic

Reflex to deletion/duplication of *ETF A, ETFB, ETFDH*

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* and *SLC37A4*

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 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 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.

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)*
