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

HERITABLE LIVER DISEASE TESTING 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 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

Reason for Testing:

- ☐ Diagnosis in symptomatic patient
- ☐ Carrier testing
- ☐ Presymptomatic diagnosis of at-risk sibling
- ☐ Prenatal diagnosis (by previous arrangement only)
- ☐ Family history of disease

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

CLINICAL HISTORY

- ☐ Jaundice
- ☐ Cholestasis
- ☐ Failure to thrive
- ☐ Characteristic facial features
- ☐ Congenital heart disease
- ☐ Vertebral body defect
- ☐ Fat malabsorption
- ☐ Rickets
- ☐ Liver cysts
- ☐ Kidney cysts
- ☐ Liver Transplant
- ☐ Elevated GTP
- ☐ Normal or low GTP
- ☐ Elevated bilirubin
- ☐ Paucity of bile ducts
- ☐ **Other Symptoms** *(Please specify):* _____

_____

TEST(S) REQUESTED

Liver Panels by Next-Generation Sequencing (NGS)

☐ **Liver Diseases Panel**

(ABCB11, ABCB4, ABCC2, ABCD3, ABCG5, ABCG8, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, CC2D2A, CFTR, CLDN1, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DHCR7, EHHADH, EPHX1, FAH, GPBAR1, HNF1A, HNF1B, HSD17B4, HSD3B7, INVS, JAG1, LIPA, MKS1, MPV17, MYO5B, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, POLG, SCP2, SERPINA1, SLC10A1, SLC10A2, SLC25A13, SLC27A5, SMPD1, TJP2, TMEM216, TRMU, UGT1A1, VIPAS39, VPS33B*

excluding exons 1, 2, and 4 in *NOTCH2* due to high homologous regions)

- ☐ Reflex to deletion/duplication of entire panel
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____

_____

Note: "Reflex to Liver Diseases Panel" can be ordered with each of the Liver Panels **below.** If the primary test results are negative or they do not fully explain the patients clinical symptoms, the Liver Diseases Panel will automatically be performed when "Reflex to Liver Diseases Panel" is also selected.

☐ **Jaundice Panel**

(ABCB4, ABCB11, ATP8B1, JAG1, TJP2)

- ☐ Reflex to deletion/duplication of entire panel
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____

_____

- ☐ Reflex to Liver Diseases Panel

☐ **Cystic Diseases of the Liver/Kidney Panel**

(ALG8, DNAJB11, DZIP1L, GANAB, LRP5, PKD2, PKHD1, PRKCSH, SEC63)

- ☐ Reflex to deletion/duplication of entire panel†
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____

_____

- ☐ Reflex to Liver Diseases Panel

☐ **Bile Acid Defects Panel**

(ABCD3, AKR1D1, AMACR, CYP7B1, HSD3B7)

- ☐ Reflex to deletion/duplication of entire panel
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____

_____

- ☐ Reflex to Liver Diseases Panel

☐ **UGT1A1 Gene Sequencing (Gilbert, Crigler-Najjar Syndromes)**

- ☐ Reflex to deletion/duplication of *UGT1A1*
- ☐ Reflex to Liver Diseases Panel

☐ **ATP7B Gene Sequencing (Wilson Disease)**

- ☐ Reflex to deletion/duplication of ATP7B
- ☐ Reflex to Liver Diseases Panel

Single Gene Testing

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

- ☐ Alagille syndrome *(JAG1)* full sequence analysis
  - ☐ Reflex to deletion/duplication of *JAG1*
- ☐ PFIC1/FIC1 deficiency *(ATP8B1)* full sequence analysis
  - ☐ Reflex to deletion/duplication of *ATP8B1*
- ☐ PFIC2/BSEP deficiency *(ABCB11)* full sequence analysis
  - ☐ Reflex to deletion/duplication of *ABCB11*
- ☐ PFIC3/MDR3 deficiency *(ABCB4)* full sequence analysis
  - ☐ Reflex to deletion/duplication of *ABCB4*
- ☐ PFIC4/Familial Hypercholanemia *(TJP2)* full sequence analysis
  - ☐ Reflex to deletion/duplication of *TJP2*
- ☐ *BAAT*/Familial Hypercholanemia full sequence analysis
  - ☐ Reflex to deletion/duplication of *BAAT*
- ☐ α1-antitrypsin deficiency *(SERPINA1)* full sequence analysis
  - ☐ Reflex to deletion/duplication of *SERPINA1*
- ☐ α1-antitrypsin *(SERPINA1)* SNP assay for PI*Z and S alleles

☐ **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 GANAB is not available at this time.

Whole Exome Sequencing
If you are interested in Whole Exome Sequencing, test requisitions are available at: www.cincinnatichildrens.org/exome

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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 <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 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 variant: _____

Patient's relation to proband: _____

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.