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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3333 Burnet Avenue, Room R1042
Cincinnati, OH 45229

MOLECULAR GENETICS 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)

☐ PATIENT BILLING

☐ Check Enclosed ☐ Money Order ☐ Credit Card (Visa, MC, Amex., Disc.)

Credit Card Number: _____

Card Holder Name: _____

Expiration Date: _____

Signature: _____

☐ REFERRING INSTITUTION

Institution: _____

Address: _____

City/State/Zip: _____

Accounts Payable Contact Name: _____

Phone: _____

Fax: _____

Email: _____

☐ INSURANCE / POLICY HOLDER INFORMATION*

Name: _____

Gender: _____ Date of Birth _____ / _____ / _____

Authorization Number: _____

Insurance Name: _____

Insurance Address: _____

City/State/Zip: _____

Insurance ID Number: _____

Group Number: _____

Insurance Phone Number: _____

* PLEASE NOTE:

1. Insurance can only be billed if requested at the time of service.
2. Acceptable forms of insurance are:
   - Commercial
   - Ohio, Indiana or Kentucky Medicaid only

   Please call 866-450-4198.

SAMPLE/SPECIMEN INFORMATION

SPECIMEN TYPE: ☐ Amniotic fluid ☐ Blood ☐ Cytobrushes

☐ 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 tissue samples.

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
- ☐ Asymptomatic infant with abnormal newborn screen
- ☐ Carrier (Heterozygote) testing
- ☐ Presymptomatic diagnosis of at-risk sibling
- ☐ Prenatal testing (by previous arrangement only)
- ☐ Family history of disease

PEDIGREE OR FAMILY HISTORY

Parental Consanguinity ☐ Y ☐ N

TEST(S) REQUESTED

Fatty Acid Oxidation Disorders

- ☐ *ACADM* (K329E) genotyping only
- ☐ *ACADM* (K329E) genotyping, with reflex to full sequencing if indicated
- ☐ *ACADM* full gene sequence analysis
- ☐ *ACADM* full gene sequence analysis, with reflex to MetaboSeq® if indicated
- ☐ *ACADVL* full gene sequence analysis
- ☐ *ACADVL* full gene sequence analysis, with reflex to MetaboSeq® if indicated
- ☐ *CPT2* full gene sequence analysis
- ☐ *CPT2* full gene sequence analysis, with reflex to MetaboSeq® if indicated
- ☐ *HADHA* (E510Q) genotype analysis
- ☐ *HADHA* (E510Q) genotype analysis, with reflex to MetaboSeq® if indicated
- ☐ MetaboSeq® (19 gene next-generation sequencing panel)
- ☐ *SLC22A5* full gene sequence analysis
- ☐ *SLC22A5* full gene sequence analysis, with reflex to MetaboSeq® if indicated

Genetic Pharmacology Services

- ☐ CCHMC psychiatry panel
- ☐ TPMT genotype analysis
- ☐ Tamoxifen 2D6 genotype analysis
- ☐ Warfarin (*CYP2C9* and *VKORC1*) genotype analysis
- ☐ Specify drug: _____ (See http://gps.cchmc.org for list)

Hearing Loss Testing

(Please provide audiogram and MRI/CT report, if available).

Hearing Loss Panels

- ☐ Hearing Loss Panel Tier I (*GJB2*, *GJB6* and 8 mitochondrial mutations)
- ☐ OtoSeq® Hearing Loss Panel (next-generation sequencing of 23 genes)
- ☐ Hearing Loss Panel Tier I with reflex to OtoSeq® Hearing Loss Panel, if indicated
- ☐ Branchiootorenal Spectrum Disorder (BOR/BOS) Panel (sequencing of *EYA1*, *SIX1*, *SIX5*)
- ☐ Branchiootorenal Spectrum Disorder (BOR/BOS) Panel with reflex to OtoSeq® reanalysis, if indicated
- ☐ Hearing loss mtDNA panel (961, 1555, 1494, 3242, 3271, 7445, 7511, 8344)
- ☐ Pendred Syndrome Panel (*SLC26A4*, *FOXI1*, *KCNJ10*)
- ☐ Pendred Syndrome Panel with reflex to OtoSeq® reanalysis, if indicated
- ☐ Usher Syndrome Panel (sequencing of *CDH23*, *CLRN1*, *GPR98*, *MYO7A*, *PCDH15*, *USH1C*, *UHS1G*, *USH2A*, *WHRN*)
- ☐ Usher Syndrome Panel with reflex to OtoSeq® reanalysis, if indicated

Single Gene Tests

- ☐ *CDH23* (*USH1D* and *DFNB12*)
- ☐ *EYA1* (Branchiootorenal spectrum disorder type 1)
- ☐ *GJB2* (connexin 26)
- ☐ *GJB6* (connexin 30) deletion analysis
- ☐ *MYO7A* (*USH1B*, DFNB2, DFNA11)
- ☐ *OTOF* (AUNB1, DFNB9)
- ☐ *SLC26A4* (Pendred syndrome, DFNB4)

Hemoglobin Testing

- ☐ Comprehensive globin gene analysis
- ☐ *HBA1* and *HBA2* (α-globin) sequence analysis
- ☐ *HBA1* and *HBA2* (α-globin) deletion analysis
- ☐ *HBB* (β-globin) sequence analysis
- ☐ *HBB* (β-globin) deletion analysis

Hereditary Immunodeficiency Testing

Autoimmune Lymphoproliferative Syndrome

- ☐ *FAS* (*TNFRSF6*)
- ☐ *FASLG* (*TNFSF6*)
- ☐ *CASP10*

☐ Somatic *FAS* sequence analysis of sorted double-negative T cell (DNTC) You MUST call 513-636-8657 or 513-636-4685 in advance for specimen requirements and to schedule this test)

Bone Marrow Failure Syndromes

- ☐ Bone Marrow Failure Syndromes Panel by next-generation sequencing (NGS)
  (MPL, RPL5, RPL11, RPL35A, RPS7, RPS10, RPS17, RPS19, RPS24, RPS26, SBDS)
- ☐ Shwachman Diamond syndrome (*SBDS*)
- ☐ **Chromosome Breakage Syndrome Panel**
  (ATM, BLM, LIG4, NBN, NHEJ1)
- ☐ **Dyskeratosis Congenita Panel**
  (DKC1, NOLA2 (NHP2), NOLA3 (NOP10), TERC (hTR), TERT, TINF2, TCAB1(WDR79, WRAP53)

Familial hemophagocytic lymphohistiocytosis

If inadequate DNA is present, we will prioritize testing according to our FHL testing algorithm, unless you indicate a different order of prioritization below.

- ___ *MUNC13-4*
- ___ *PRF1*
- ___ *RAB27A* (Griscelli syndrome)
- ___ *STXBP2*
- ___ *STX11*

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TEST(S) REQUESTED CONTINUED

Hereditary Immunodeficiency Testing continued

Fanconi anemia

- ☐ Fanconi Anemia Panel by next-generation sequencing (NGS) *(FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ, FANCL, FANCM, FANCN, FANCO)*
- ☐ *FANCA*
- ☐ *FANCC*
- ☐ *FANCC* IVS4+4 A>T (common Ashkenazi mutation) only
- ☐ *FANCG*

Lymphoproliferative disorders (Including EBV-Related)

- ☐ *SH2D1A*
- ☐ *XIAP/BIRC4*
- ☐ *ITK*
- ☐ *MAGT1*

Severe Combined Immunodeficiencies

- ☐ Severe Combined Immunodeficiency Panel by next-generation sequencing (NGS) *(ADA, CD3D, CD3E, DCLRE1C, FOXN1 (WHN), IL2RG, IL7R, JAK3, LIG4, NHEJ1, ORAI1, PNP, PTPRC, RAG1, RAG2, RMRP, STAT5B, STIM1, TBX1, ZAP70)*
- ☐ X-linked severe combined immunodeficiency (*IL2RG*)

Severe congenital neutropenia

- ☐ *ELA2* (*ELANE*)
- ☐ *HAX1*
- ☐ *WAS* (males only)

☐ IPEX syndrome (*FOXP3*)

☐ Wiskott-Aldrich syndrome (*WAS*)

☐ X-linked hyper IgM immunodeficiency (*CD40LG*)

Hereditary Liver Disease Testing

☐ JaundiceChip re-sequencing array

OR

☐ *ABCB4* (PFIC3/MDR3 deficiency)
☐ *ABCB11* (PFIC2/BSEP deficiency)
☐ *ATP8B1* (PFIC1/FIC1 deficiency)
☐ *JAG1* (Alagille syndrome)
☐ *SERPINA1* genotyping assay for PI*Z and PI*S alleles
☐ *SERPINA1* (alpha-1-antitrypsin deficiency)

Intellectual Disabilities Testing

Creatine deficiency syndromes

- ☐ *SLC6A8* (creatine transporter defect)
- ☐ *GATM* (AGAT deficiency)
- ☐ *GAMT*

☐ Fragile X syndrome (*FMR1* – By PCR and Southern)
☐ *MECP2* (Rett syndrome)
☐ Prader-Willi/Angelman syndrome (by methylation specific PCR)

Lysosomal Storage Disease Testing

Cystinosis

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

Fabry Disease

- ☐ *GLA* full gene sequencing

Gaucher Disease

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

MPS II - Hunter syndrome

- ☐ *IDS* full gene sequencing

Pompe Disease

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

Mitochondrial Disorders

Mitochondrial DNA

- ☐ Whole mitochondrial genome (mtDNA) sequencing
- ☐ mtDNA large deletions/duplications

mtDNA panels

- ☐ Common mutations mtDNA panel (mtDNA 1555, 3243, 3271, 3460, 8344, 8993, 11778, 14459, 14484)
- ☐ Common mutations mtDNA panel with reflex to mtDNA whole genome sequencing

mtDNA panels continued

- ☐ Neuromuscular disorders mtDNA panel (MELAS/MERRF: mtDNA 3243, 3271, 8344)
- ☐ Neuromuscular disorders mtDNA panel with reflex to mtDNA whole genome sequencing
- ☐ Leber Hereditary Optic Neuropathy mtDNA panel (mtDNA 3460, 11788, 14459, 14484)
- ☐ Leber Hereditary Optic Neuropathy mtDNA panel with reflex to mtDNA whole genome sequencing
- ☐ Hearing loss mtDNA panel (mtDNA 1555, 1494, 961, 7445, 7511)

POLG-related disorders: AD-PEO, SANDO, MIRAS

- ☐ *POLG1* full gene sequencing

IBMPFD

- ☐ *VCP* full gene sequencing

Oncology Molecular Testing

Samples must be received within 24 hours of drawing.

- ☐ BCR/ABL t(9;22)--(qualitative only)
- ☐ BCR/ABL (quantitative p210)
- ☐ *JAK2*
- ☐ PML-RARα t(15;17)—(qualitative only)
- ☐ *NPM1* (quantitative)

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TEST(S) REQUESTED CONTINUED

Thrombophilia Testing

☐ Factor V (Leiden)

☐ *MTHFR* (677 C>T and 1298 A>C) genotype

☐ Prothrombin (Factor II) G20210A genotype

☐ Thrombophilic polymorphism panel (*MTHFR* 677C>T and 1298 A>C, Factor V- Leiden, Factor II-Prothrombin G20210A, PAI-1 4G/5G genotypes)

Other Services

☐ DNA/RNA processing and storage (Call (513) 636-4474 to arrange)

☐ Maternal cell contamination (by STR; required for all prenatal diagnostic testing)

☐ Twin zygosity (by STR)

Other Testing

☐Bone marrow engraftment by STR - same sex donor and recipient

Please specify:

☐ Donor ☐ Host (pre-transplant) ☐ Host (post-transplant)

Special study: Bone marrow engraftment

(WBC sub-populations) ☐ STR ☐ FISH

You must call 513-636-8657 or 513-636-4685 to schedule this test prior to sample submission.

☐ Hereditary hemochromatosis (HFE- C282Y and H63D)

☐ **Targeted (family specific) mutation analysis**

Gene of interest _____

Proband's name _____

Proband's DOB _____

Proband's mutation _____

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