

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)

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

Has patient received a bone marrow transplant? Yes No

If yes, date of bone marrow transplant _____

Percent engraftment _____

Please send saliva kit and two cytobrushes. Note: STR analysis at an additional charge is required on cytobrushes and saliva samples obtained on all patients post BMT.

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

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.

REFERRING PHYSICIAN

Physician Name (print): _____

Address: _____

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

Email: _____

Genetic Counselor/Lab Contact Name: _____

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

Email: _____

_____ Date: ____/____/____

Referring Physician Signature (REQUIRED)

INDICATIONS/DIAGNOSIS/ICD-10 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

SAMPLE/SPECIMEN INFORMATION

Has patient received a bone marrow transplant?

- Yes
- No

If yes, date of bone marrow transplant: _____

Percent engraftment: _____

PEDIGREE OR FAMILY HISTORY

Parental Consanguinity Y N

TEST(S) REQUESTED

Custom Gene Testing

Sanger sequencing of clinically relevant rare disease genes is available by prior arrangement. Custom single gene requisition available at: www.cincinnatichildrens.com/customtesting

Deletion/Duplication Assay

Custom deletion/duplication analysis available for many genes. List of available genes and test requisition available at: www.cincinnatichildrens.com/deldup

Whole Exome Sequencing

Exome sequencing test requisition available at: www.cincinnatichildrens.org/exome

Epidermolysis Bullosa

- EBSeq Epidermolysis Bullosa Panel
(*CD151, CDSN, CHST8, COL17A1, COL7A1, DSP, DST, EXPH5, FERMT1 (KIND1), ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT5, KRT14, LAMA3, LAMB3, LAMC2, PKP1, PLEC1, TGM5*)

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*

- Psychiatry Pharmacogenetics Expanded Panel
- TPMT Genotype Analysis
- Opioid CYP2D6 Pharmacogenetics Panel
- CYP2C19
- CYP2C9
- CYP2D6
- CYP2D6/CYP2C19
- CYP3A5 (if liver transplant, must test DONOR sample)
- Tamoxifen
- Warfarin

*DONOR sample required for liver transplant patients

Hearing Loss Testing

Hearing Loss Panels

(Provide audiogram and MRI/CT report)

- Hearing Loss Panel Tier I (*GJB2* sequencing, *GJB6* deletion analysis 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, FOX11, KCNJ10*)
- Pendred Syndrome Panel with reflex to OtoSeq[®] reanalysis, if indicated
- Usher Syndrome Panel (sequencing of *CDH23, CLRN1, GPR98, MYO7A, PCDH15, USH1C, USH1G, 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*)

TEST(S) REQUESTED CONTINUED

Hematology Testing

- Dense Deposit Disease/C3 Glomerulonephritis Sequencing Panel**
(Includes C3, CD46 (MCP), CFB, CFD, CFH, CFHR2, CFHR5 and CFI)

Hemoglobin Testing

- Alpha (HBA1/2) and Beta (HBB) Globin Gene Locus Analysis
 - HBA1 and HBA2 (α-globin) sequence analysis
 - HBA1 and HBA2 (α-globin) locus del/dup analysis (HBA1/2 & HBZ)
 - HBB (β-globin) sequence analysis
 - HBB (β-globin) locus del/dup analysis (HBB, HBD, HBG1/2, & HBE)
- Hemoglobin D (HBD) Delta (HBD) Globin Locus Sequence Analysis
- Gamma (HBG1/2) Globin Gene Analysis by Custom Gene Sequencing

Hemolytic Anemia

- Hemolytic Anemia Panel (includes sequence analysis of ABCG5, ABCG8, AK1, ALAS2, ALDOA, ANK1, ATP11C, C15orf41, CDAN1, COL4A1, EPB41, EPB42, G6PD, GATA1, GCLC, GPI, GPX1, GSR, GSS, GYPC, HK1, KCNN4, KIF23, KLF1, LPIN2, NT5C3A, PFKM, PGK1, PIEZO1, PKLR, RHAG, SEC23B, SLC2A1 (GLUT1), SLC4A1, SPTA1, SPTB, TPI1, XK)
- Congenital Dyserythropoietic Anemia (CDA) Panel (includes sequence analysis of ALAS2, C15orf41, CDAN1, GATA1, KIF23, KLF1, LPIN2, SEC23B)
- CDA Panel with reflex to Hemolytic Anemia Panel reanalysis, if indicated
- RBC Membrane Disorders Panel (includes sequence analysis of ABCG5, ABCG8, ANK1, ATP11C, COL4A1, EPB41, EPB42, GYPC, KCNN4, PIEZO1, RHAG, SLC2A1, SLC4A1, SPTA1, SPTB, XK)
- RBC Membrane Disorders Panel with reflex to Hemolytic Anemia Panel reanalysis, if indicated
- RBC Enzymopathy Panel (includes sequence analysis of AK1, ALDOA, G6PD, GCLC, GPI, GPX1, GSR, GSS, HK1, NT5C3A, PFKM, PGK1, PKLR, TPI1)
- RBC Enzymopathy Panel with reflex to Hemolytic Anemia Panel reanalysis, if indicated

Thrombophilia Testing

- Factor V (Leiden)
- MTHFR (677 C>T and 1298 A>C) genotype
- Prothrombin (Factor II) G20210A genotype
- Thrombophilic polymorphism panel (Factor V- Leiden and Factor II-Prothrombin G20210A)

Thrombotic Microangiopathy (aHUS and TTP)

- aHUS Genetic Susceptibility Panel (Includes sequence analysis of ADAMTS13, C3, C4BPA, CD46 (MCP), CD59, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, PLG, THBD and deletion/duplication analysis of CFHR1 and CFHR3 via MLPA. Also includes analysis of variants c.2653C>T and c.2654G>A in the C5 gene, which are associated with poor response to eculizumab)
- ADAMTS13 gene sequencing

Hereditary Immunodeficiency Testing

Autoimmune Lymphoproliferative Syndrome

- Autoimmune Lymphoproliferative Syndrome (ALPS) Panel by next generation sequencing (NGS)
(ADA2 (CECR1), CASP10, CASP8, CTLA4, FADD, FAS, FASLG, ITK, KRAS, LRBA, MAGT1, NRAS, PRKCD, RASGRP1, STAT3)
- FAS (TNFRSF6)
- FASLG (TNFSF6)
- CASP10

Somatic FAS sequence analysis of sorted double-negative T cell (DN2C)
You MUST call 513-636-2731 in advance for specimen requirements and to schedule this test!

Bone Marrow Failure Syndromes Panel by next-generation sequencing (NGS)

(ABCB7, ACD, ADA2 (CECR1), AK2, AP3B1, ATM, ATR, BLM, BRCA1, BRCA2, BRIP1, CD40LG, CLPB, CSF3R, CTC1, CXCR2, CXCR4, DKC1, DNAJC21, EFL1, EIF2AK3, ELANE, EPO, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GF11, HAX1, HYOU1, JAGN1, LAMTOR2, LIG4, LYST, MAD2L2, MPL, MRTFA (MKL1), MYSM1, NAF1, NBN, NHEJ1, NHP2, NOP10, NSMCE3, PALB2, PARN, POT1, RAB27A, RAC2, RAD51, RAD51C, RBM8A, RFWDD3, RMRP, RNF168, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27a, RPS28, RPS29, RPS7, RTEL1, RUNX1, SBDS, SLC37A4, SLX4, SMARCD2 (except exon1), SRP54, SRP72, STK4, STN1, TAZ, TCIRG1, TCN2, TERC, TERF2IP, TERT, TINF2, TP53, TSR2, UBE2T, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XRCC2)

Chromosome Breakage Syndrome Panel

(ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, LIG4, MAD2L2, MYSM1, NBN, NHEJ1, NSMCE3, PALB2, RAD51, RAD51C, RFWDD3, SLX4, UBE2T, XRCC2)

Diamond-Blackfan anemia panel

(EPO, GATA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27a, RPS28, RPS29, RPS7, TSR2)

Dyskeratosis congenita Panel

(ACD, CTC1, DKC1, NAF1, NHP2, NOP10, PARN, POT1, RTEL1, STN1, TERC, TERF2IP, TERT, TINF2, WRAP53)

Familial hemophagocytic lymphohistiocytosis

- Hemophagocytic Lymphohistiocytosis (HLH) Panel
(AP3B1, AP3D1 (except exon 23), CD27, CD70, CTPS1, GATA2, ITK, LYST, MAGT1, NLRC4, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D, XIAP)

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

- ___ UNC13D (MUNC13-4)
- ___ PRF1
- ___ RAB27A (Griscelli syndrome)
- ___ STXBP2
- ___ STX11

Fanconi anemia

- Fanconi Anemia Panel by next-generation sequencing (NGS)
(BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFWDD3, SLX4, UBE2T, XRCC2)
- FANCA

Lymphoproliferative disorders (Including EBV-Related)

- SH2D1A
- XIAP (BIRC4)
- ITK
- MAGT1

TEST(S) REQUESTED CONTINUED

Severe Combined Immunodeficiencies

- Severe Combined Immunodeficiency Panel by next-generation sequencing (NGS)
(ADA, AK2, ATM, BCL11B, CD247, CD3D, CD3E, CDH17, CHD7, CIITA, CORO1A, DCLRE1C, DOCK8, FOXP1, IL2RG, IL7R, JAK3, LAT, LCK, LIG4, MSN, NHEJ1, ORAI1, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RMRP, STAT5B, STIM1, STK4, TAP1, TAP2, TBX1, TTC7A, ZAP70)
- X-linked severe combined immunodeficiency (*IL2RG*)

Severe congenital neutropenia

- Inherited Neutropenia Panel by next-generation sequencing (NGS)
(AK2, AP3B1, CD40LG, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, EIF2AK3, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, HYOU1, JAGN1, LAMTOR2, LYST, MRTFA (MKL1), RAB27A, RAC2, RMRP, RUNX1, SBDS, SLC37A4, SMARCD2 (except exon1), SRP54, STK4, TAZ, TCIRG1, TCN2, TP53, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1)
- ELANE (ELA2)*
- HAX1*
- WAS (males only)*

Other Primary Immunodeficiencies

- IPEX syndrome (*FOXP3*)
- Shwachman Diamond syndrome (*SBDS*)
- Wiskott-Aldrich syndrome (*WAS*)
- X-linked hyper IgM immunodeficiency (*CD40LG*)

Hereditary Liver Disease Testing

- 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*)
- Jaundice Panel (*ABCB4, ABCB11, ATP8B1, JAG1, TJP2*)
- Cystic Diseases of the Liver/Kidney Panel (*ALG8, DNAJB11, DZIP1L, GANAB, LRP5, PKD2, PKHD1, PRKCSH, SEC63*)
- Bile Acid Defects Panel (*ABCD3, AKR1D1, AMACR, CYP7B1, HSD3B7*)

Heritable Pancreatic Disease Testing

- Pancreas Panel (*CASR, CEL, CFTR, CLDN2, CPA1, CTSC, PRSS1, SBDS, SPINK1, UBR1*)
- Pancreatic Insufficiency Panel (*CEL, CFTR, SBDS, UBR1*)
- SBDS* full sequence analysis

Single Gene Tests

- ABCB4* (PFIC3/MDR3 deficiency)
- ABCB11* (PFIC2/BSEP deficiency)
- ATP7B* Gene Sequencing (Wilson Disease)
- ATP8B1* (PFIC1/FIC1 deficiency)
- BAAT* (Familial Hypercholanemia)
- JAG1* (Alagille syndrome)
- SERPINA1* genotyping assay for PI*Z and PI*S alleles
- SERPINA1* (alpha-1-antitrypsin deficiency)
- TJP2* (PFIC4/Familial Hypercholanemia)
- UGT1A1* Gene Sequencing (Gilbert, Crigler-Najjar Syndromes)

Intellectual Disabilities Testing

Creatine deficiency syndromes

- SLC6A8* (creatine transporter defect)
- GATM* (AGAT deficiency)
- GAMT*
- Fragile X syndrome (*FMR1* – By PCR and mPCR)
- MECP2* sequencing (Rett syndrome)
- MECP2* deletion/duplication analysis by MLPA
- Prader-Willi/Angelman syndrome (by MLPA)

Lysosomal Storage Disease Testing

Cystinosis

- CTNS* full gene sequencing only
- CTNS* 57 kb deletion analysis by MLPA only
- 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

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

- POLG* 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
- BCR/ABL t(9;22) Quantitative (p210)
- BCR/ABL t(9;22) Quantitative (p190)
- PML-RAR α t(15;17) – (qualitative only)
- JAK2* QUANTITATIVE (V617F)

Oncology Molecular Testing *Samples must be received within 72 hours of drawing.*

- BRAF* full gene sequence analysis
- BRAF* (V600E) mutation only – **Does patient have LCH?** Yes No
- BRAF* V600E by real-time PCR (no FFPE tissue accepted)

Targeted (family specific) mutation analysis

Gene of interest: _____
 Proband's name: _____
 Proband's DOB (mm/dd/yyyy): _____
 Proband's variant: _____
 Relationship to proband: _____

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

- Bone marrow engraftment by STR - same sex donor and recipient
Please specify: Donor Host (pre-transplant) Host (post-transplant)
- Sorted cell engraftment study (WBC sub-population)
You must call 513-636-4474 to schedule this test prior to sample submission.
Please specify: STR FISH
- Hereditary hemochromatosis (HFE- C282Y and H63D)
- DNA/RNA processing and storage (Call (513) 636-4474 to arrange)
- Maternal cell contamination (by STR; required for all prenatal diagnostic testing)
- Maternal Engraftment
- Twin zygosity (by STR)