Cincinnati Children's
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GENETICS AND GENOMICS DIAGNOSTIC LABORATORY
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

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 _____ / _____ / _____

Sex Assigned at Birth: ☐ Male ☐ Female ☐ Uncertain/Other: _____

ETHNIC/RACIAL BACKGROUND (Choose all that apply)

☐ 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. Please contact the lab at 513-636-4474 for a free saliva kit.

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

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 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-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.org/customtesting

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

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

EPIDERMOLYSIS BULLOSA TESTING

- ☐ 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)

GENETIC PHARMACOLOGY SERVICES TESTING*

- ☐ Comprehensive Pharmacogenetics Panel
- ☐ CYP2C19
- ☐ CYP2C9
- ☐ CYP2D6
- ☐ CYP2D6/CYP2C19 (with or without BMT patient interp)
- ☐ CYP3A5
- ☐ Opioid CYP2D6 Pharmacogenetics Panel
- ☐ Psychiatry Pharmacogenetics Expanded Panel
- ☐ TPMT/NUDT15 Genotype Analysis (Thiopurine Panel)
- ☐ 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)
- ☐ Hearing Loss Panel Tier I with reflex to OtoSeq Hearing Loss Panel*
- ☐ OtoSeq Hearing Loss Panel
  (ADGRV1, CDH23, CLRN1, EYA1, FOXI1, GJB2, GJB6, KCNJ10, MYO6, MYO7A, OTOF, PCDH15, POU3F4, SIX1, SIX5, SLC26A4, TMC1, TMIE, TMPRSS3, USH1C, USH1G, USH2A, WHRN)
- ☐ Branchiootorenal Spectrum Disorder (BOR/BOS) Panel *(EYA1, SIX1, SIX5)*
- ☐ Branchiootorenal Spectrum Disorder (BOR/BOS) Panel with reflex to OtoSeq reanalysis*
- ☐ 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*
- ☐ Usher Syndrome Panel
  (*CDH23*, *CLRN1*, *GPR98*, *MYO7A*, *PCDH15*, *USH1C*, *USH1G*, *USH2A*, *WHRN*)
- ☐ Usher Syndrome Panel with reflex to OtoSeq® reanalysis*

Hearing Loss 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)

HEMATOLOGY TESTING

Dense Deposit Disease Testing

- ☐ **Dense Deposit Disease/C3 Glomerulonephritis Sequencing Panel**
  (*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 Testing

- ☐ Hemolytic Anemia Panel
  (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
  (ALAS2, C15orf41, CDAN1, GATA1, KIF23, KLF1, LPIN2, SEC23B)
- ☐ CDA Panel with reflex to Hemolytic Anemia Panel reanalysis*
- ☐ RBC Membrane Disorders Panel
  (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

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Patient Name: _____ Date of Birth: _____

TEST(S) REQUESTED CONTINUED

☐ RBC Enzymopathy Panel
(AK1, ALDOA, G6PD, GCLC, GPI, GPX1, GSR, GSS, HK1, NT5C3A, PFKM, PGK1, PKLR, TPI1)

☐ RBC Enzymopathy Panel with reflex to Hemolytic Anemia Panel reanalysis*

☐ **Platelet Disorders Gene Sequencing Panel**
Platelet Disorders Gene Sequencing Panel requisition must be completed and is available at: www.cincinnatichildrens.org/service/g/genetics-genomics-diagnostic-lab/requisition
(ABCG5, ABCG8, ACBD5, ACTN1, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S6, CD36, CYCS, DIAPH1, DTNBP1, ETV6, FERMT3, FLI1, FLNA, FYB1, GATA1, GFI1B, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB3, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEA, NBEAL2, ORAI1, P2RX1, P2RY1, P2RY12, PLA2G4A, PRKACG, PTGS1, RAB27A, RASGRP2, RBM8A, RUNX1, SLFN14, STIM1, STX11, STXBP2, TBXA2R, TBXAS1, THPO, TUBB1, UNC13D, VIPAS39, VPS33B, VPS45, WAS)

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
(*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

Additional Panels

☐ **Erythrocytosis Gene Sequencing Panel**
(BHLHE41, BPGM, CALR, CYB5R3, EGLN1 (PHD2), EGLN2 (PHD1), EGLN3 (PHD3), EPAS1, EPO, EPOR, GFI1B, HBA1, HBA2, HBB, HIF1A, HIF1AN (FIH), HIF3A, JAK2, KDM6A, MPL, OS9, PIEZO1, PKLR, SH2B3, SLC30A10, VHL, ZNF197)

☐ **Thrombocytosis Gene Sequencing Panel**
(CALR, JAK2, MPL, THPO)

HEREDITARY IMMUNODEFICIENCY TESTING

Autoimmune Lymphoproliferative Syndrome

☐ Autoimmune Lymphoproliferative Syndrome (ALPS) Gene Seq Panel
(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 (DNTC)
You MUST call 513-636-2731 in advance for specimen requirements and to schedule this test

☐ **Bone Marrow Failure Syndromes Gene Sequencing Panel**
(ABCB7, ACD, ADA2 (CECR1), AK2, ALAS2, ANKRD26, AP3B1, ATM, ATR, BLM, BRCA1, BRCA2, BRIP1, C15orf41, CARD11, CBL, CD40LG, CDAN1, CEBPA, CLPB, CSF3R, CTC1, CXCR2, CXCR4, CYCS, DDX41, DKC1, DNAJC21, DNMT3A, DUT, EFL1, EIF2AK3, ELANE, EPO, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, GLRX5, GP1BA, GP1BB, GP9, GRHL2, HAX1, HOXA11, HYOU1, IKZF1, ITGA2B, ITGB3, JAGN1, JAK2, KIF23, KIT, KLF1, KRAS, LAMTOR2, LIG4, LYST, MAD2L2, MASTL, MBD4, MECOM, MPL, MRTFA (MKL1), MYH9, MYSM1, NAF1, NBN, NHEJ1, NHP2, NOP10, NSMCE3, PALB2, PARN, PAX5, PGM3, POT1, PTPN11, PUS1, RAB27A, RAC2, RAD51, RAD51C, RBM8A, RFWD3, RMRP, RNF168, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, RUNX1, SALL4, SAMD9, SAMD9L, SBDS, SBF2, SEC23B, SH2B3, SLC19A2, SLC25A38, SLC35C1, SLC37A4, SLX4, SMARCD2, SRP54, SRP72, STIM1, STK4, STN1, TAZ, TCIRG1, TCN2, TERC, TERF2IP, TERT, TET2, THPO, TINF2, TLR8, TP53, TRNT1, TSR2, TUBB1, UBE2T, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XRCC2, YARS2, ZCCHC8)

☐ **Chromosome Breakage Disorders Panel**
(ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, LIG4, MAD2L2, MYSM1, NBN, NHEJ1, NSMCE3, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2)

☐ **Diamond-Blackfan anemia panel**
(EPO, GATA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15, RPS15A, 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)

☐ **Immunology Exome**
Gene sequencing panel with 442 genes associated with immune system defects or related heritable disorders. Primary Immunodeficiency requisition must be completed and is available at: www.cincinnatichildrens.org/service/g/genetics-genomics-diagnostic-lab/requisition

Familial hemophagocytic lymphohistiocytosis

☐ Hemophagocytic Lymphohistiocytosis (HLH) Panel
(AP3B1, AP3D1 (except exon 23), CD27, CD70, CDC42, CTPS1, CYBA, CYBB, CYBC1, GATA2, ITK, LYST, MAGT1, NCF2, NCF4, NLRC4, PRF1, RAB27A, RASGPR1, RC3H1, RHOG, 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, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2)

☐ *FANCA*

Lymphoproliferative disorders (Including EBV-Related)

☐ *SH2D1A*
☐ *XIAP (BIRC4)*
☐ *ITK*
☐ *MAGT1*

Severe Combined Immunodeficiencies (SCID)

☐ Severe Combined Immunodeficiency (SCID) Gene Sequencing Panel
(ADA, AK2, ATM, BCL11B, CD247, CD3D, CD3E, CDH17, CHD7, CIITA, CORO1A, DCLRE1C, DOCK8, FOXN1, 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 Gene Sequencing Panel
(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)*

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Patient Name: _____ Date of Birth: _____

TEST(S) REQUESTED CONTINUED

HEREDITARY LIVER DISEASE TESTING

☐ Liver Diseases Panel
(ABCB11, ABCB4, ABCC12, 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 *(ABCB11, ABCB4, 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)*

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)

HERITABLE PANCREATIC DISEASE TESTING

☐ Pancreas Panel
(CASR, CEL, CFTR, CLDN2, CPA1, CTRC, PRSS1, SBDS, SPINK1, UBR1 with PRSS1 deletion/duplication via MLPA)

☐ Pancreatic Insufficiency Panel *(CEL, CFTR, SBDS, UBR1)*

☐ *SBDS* full sequence analysis

INBORN ERRORS OF METABOLISM

☐ MetaboSeq Gene Sequencing Panel
(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)

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

☐ Riboflavin Disorders Gene Sequencing Panel
(ETFA, ETFB, ETFDH, SLC52A2, SLC52A3)

☐ Elevated C16 Gene Sequencing Panel
(SLC25A20, CPT2)

☐ L CHAD/TFP Gene Sequencing Panel for Long Chain 3 Hydroxyacyl CoA Dehydrogenase (LCHAD) Deficiency/Trifunctional Protein Deficiency (TFP)
(HADHA, HADHB)

☐ GSD type I Gene Sequencing Panel
(G6PC, SLC37A4)

Single Gene Sequencing

☐ *ACADM* (K329E) genotyping only
☐ *ACADM* full gene sequence analysis
☐ *ACADVL* full gene sequence analysis
☐ *ALDOB* full gene sequence analysis
☐ *CPT2* full gene sequencing analysis
☐ *GAA* full gene sequence analysis
☐ *SLC22A5* full gene sequencing analysis

INTELLECTUAL DISABILITIES TESTING

☐ Autism, Intellectual Disability, and Developmental Delay Panel
Gene sequencing panel with more than 2,750 genes associated with autism spectrum disorder, intellectual disability and/or developmental delay. Autism, Intellectual Disability and Developmental Delay Gene Sequencing Panel Requisition must be completed and is available at: www.cincinnatichildrens.org/service/g/genetics-genomics-diagnostic-lab/requisition

☐ Rubinstein-Taybi and Related Syndromes Gene Panel
(CREBBP, EP300, HNRNPH1, HNRNPH2, SIN3A, SIN3B, SRCAP and copy number analysis of *CREBBP* and *EP300)*

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

Fabry Disease

☐ *GLA* full gene sequencing

Gaucher Disease

☐ *GBA* full gene sequencing

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

HEARING LOSS

☐ Hearing loss mtDNA panel
(961, 1555, 1494, 3242, 3271, 7445, 7511, 8344)

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-RARα t(15;17)—(qualitative only)
☐ 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

Samples must be received within 48 hours of drawing.

☐ Bone marrow engraftment by STR - same sex donor and recipient
Please specify: ☐ Donor ☐ Host (pre-transplant) ☐ Host (post-transplant)
☐ *JAK2* Quantitative (V617F)
☐ *MAP2K1* full gene sequencing analysis (**% Tumor**:_____)
☐ *PTEN* sequencing

Cincinnati Children's
changing the outcome together

Patient Name: _____ Date of Birth: _____

TEST(S) REQUESTED CONTINUED

ADDITIONAL MOLECULAR TESTING

☐ *ABCD1* gene sequencing (X-Linked Adrenoleukodystrophy)

☐ Reflex to *ABCD1* deletion/duplication analysis by MLPA

☐ *ABCD1* deletion/duplication analysis by MLPA

☐ **Circadian and Complex Sleep Disorders Gene Sequencing Panel**
(ADCY3, ADK, ADORA2A, ADRB1, AK5, APP, ARNTL, ARNTL2, ATP2B3, BDNF, BHLHE40, BHLHE41, BLOC1S6, BTBD9, CACNA1A, CACNA1B, CACNA1G, CAMK2A, CAMK2B, CAMTA1, CDKL5, CHRM1, CHRM3, CIART, CLOCK, CNTNAP2, CREB1, CREBBP, CRH, CRY1, CRY2, CSNK1A1, CSNK1D, CSNK1E, CUL3, DBH, DBP, DISC1, EGR3, ELP3, ERC2, FAAH, FABP7, FBXL3, FMR1, FOS, FOSB, FOXP1, FTO, FUS, GRIA1, GRIA3, GRIN1, GRM1, GRM2, GRM3, HCRT, HCRTR2, HDC, HLF, HOMER1, HOMER2, HTR1A, HTR1B, HTR2A, HTR2C, HTR7, HTT, IFNAR1, IL1R1, IL6, JAML, KANSL1, KCNA2, KCNA3, KCNC1, KCNK9, KCNN3, KCTD5, KPNB1, LEP, MAP2K5, MCHR1, MEIS1, MTOR, NALCN, NCKAP5, NFKB1, NLGN2, NLGN3, NLRP3, NOS1, NPAS2, NPRL3, NPSR1, NR1D1, NR1D2, NTSR1, OPN4, OPRM1, PANX1, PAX8, PCDHA3, PDE4D, PER1, PER2, PER3, PPARGC1A, PPP3CA, PPP3R1, PRKAB2, PRKG1, PRL, PRNP, PROK2, PTPA, PTPRD, RAB3A, RCAN2, RGS16, RIMS1, RORA, RORB, RORC, SCN1A, SHANK3, SHMT1, SIK3, SLC18A2, SLC29A1, SLC6A2, SLC6A3, SLC6A4, TEF, TIMELESS, TNF, TNFRSF1A, TNRC6B, TOX3, TRANK1, UBB, UBE3A, VAMP2)

☐ **Cleft and Craniofacial Gene Panel (288 genes)**
ABCC9, ACSS2, ACTB, ACTG1, ADAMTSL4, AHDC1, ALPL, ALX1, ALX3, ALX4, AMELX, AMER1, AMMECR1, AMOTL1, ANKH, ANKRD11, ARHGAP29, ARSB, ASPH, ASXL1, ASXL3, B3GAT3, B3GLCT, BCOR, BMP2, BMP4, BMPR1B, BPNT2, BRAF, BRD4, C2CD3, CBFB, CCNQ, CD96, CDC45, CDH1, CDKN1C, CDON, CENPF, CEP164, CHD5, CHD7, CILK1, CNOT1, COG1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, CPLANE1, CREBBP, CTNND1, CTSK, CYP26B1, DDX59, DHCR7, DHODH, DISP1, DLL1, DLX4, DPF2, DPH1, DVL1, DVL3, EDN1, EDNRA, EFNA4, EFNB1, EFTUD2, EHMT1, EIF4A3, EP300, ERF, ESCO2, ESRP2, EVC, EVC2, EYA1, FAM20C, FBN1, FGD1, FGF10, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FOXE1, FOXI3, FRAS1, FREM1, FST, FTO, FZD2, GAS1, GDF11, GJA1, GLI2, GLI3, GNAI3, GNAS, GNPTAB, GPC3, GPC4, GRHL3, GSC, GTF2E2, GZF1, HDAC8, HIST1H1E, HNRNPK, HUWE1, HYAL2, HYLS1, IDS, IDUA, IFT122, IFT140, IFT43, IGF1R, IGF2, IHH, IL11RA, INPPL1, IRF6, IRX5, ISM1, JAG1, KAT6A, KAT6B, KDM1A, KDM6A, KIAA0586, KIF7, KMT2D, KRAS, LOXL3, LRP2, LTBP1, MAFB, MAP3K7, MASP1, MED13L, MED25, MEGF8, MEIS2, MID1, MKS1, MN1, MSX1, MSX2, MTX2, MYCN, MYMK, MYT1, NBAS, NECTIN1, NEDD4L, NIPBL, OFD1, P4HB, PAX1, PAX3, PAX7, PDE4D, PGM1, PHEX, PHF21A, PHF8, PIEZO2, PIGN, PJA1, PLCB4, PLCH1, PLEKHA5, PLEKHA7, PLOD3, POLR1A, POLR1B, POLR1C, POLR1D, POR, PORCN, PPP1R12A, PRRX1, PSAT1, PTCH1, PTDSS1, PTPN11, RAB23, RAD21, RAX, RBM10, RECQL4, RIPK4, ROR2, RPGRIP1L, RPL5, RSPRY1, RUNX2, RYK, SATB2, SCARF2, SCLT1, SCN4A, SEC24D, SEMA3E, SF3B2, SF3B4, SHH, SHOC2, SHROOM3, SIN3A, SIX1, SIX2, SIX3, SIX5, SKI, SLC25A24, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCB1, SMC1A, SMC3, SMG9, SMO, SMS, SMURF1, SNRPB, SON, SOST, SOX11, SOX6, SOX9, SPECC1L, SPRY1, SPRY4, STAG2, STIL, SUFU, SUMO1, TBC1D32, TBX1, TBX22, TCF12, TCOF1, TFAP2A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TLK2, TMCO1, TOPORS, TP63, TRAF7, TRRAP, TWIST1, TWIST2, TXNL4A, UBE3B, USP9X, VAX1, VCAN, WASHC5, WDR19, WDR35, WNT5A, YAP1, YWHAE, ZEB2, ZIC1, ZIC2, ZNF462, ZSWIM6

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

☐ **Hereditary hemochromatosis (HFE- C282Y and H63D)**

☐ Maternal Cell Contamination (by STR; required for all prenatal diagnostic testing)

☐ Maternal Engraftment

☐ **Neurovascular Diseases and Stroke Gene Panel**
Neurovascular Diseases and Stroke Gene Panel requisition must be completed and is available at: www.cincinnatichildrens.org/service/g/genetics-genomics-diagnostic-lab/requisition
(ABCC6, ACTA2, ACVRL1, ADA2 (CECR1), ATP1A2, ATP7A, ATR, BRAF, CACNA1A, CBS, CCM2, CENPJ, CEP152, CEP63, CHD4, CLDN14, CNOT3, COL3A1, COL4A1, COL4A2, COLGALT1, EFNB2, ENG, EPHA4, EPHB4, FBN1, G6PC, GDF2, GLA, GUCY1A3, HBB, HRAS, HTRA1, JAG1, KRAS, KRIT1, MAP2K1, MYH11, MYLK, NF1, NHLRC2, NIN, NOTCH2, NOTCH3, NRAS, OTC, P2RY1, P2RY12, PCNT, PDCD10, PMM2, POLG, PRRT2, PTPN11, RAF1, RASA1, RBBP8, RNF213, SAMHD1, SCN1A, SCN5A, SETD5, SLC19A2, SLC2A10, SMAD2, SMAD3, SMAD4, SMARCAL1, SOS1, SUOX, TGFB2, TGFB3, TGFBR1, TGFBR2, TREX1, TSC1, TSC2, TTC19, WFS1, YY1AP1)

☐ *SMN1/SMN2* Copy Number Analysis (Spinal Muscular Atrophy)

☐ **Stickler Syndrome Gene Panel (13 genes)**
BMP4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, GZF1, LOXL3, LRP2, PLOD3, SOX9, VCAN

☐ **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.

☐ **Treacher Collins Syndrome and Mandibulofacial Dysostosis Gene Panel (10 genes)**
DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D, SF3B4, TCOF1, TXNL4A

☐ Twin zygosity (by STR)