

## 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.

## 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](http://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](http://www.cincinnatichildrens.org/deldup)

### Whole Exome Sequencing

Exome sequencing test requisition available at: [www.cincinnatichildrens.org/exome](http://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\*

## TEST(S) REQUESTED CONTINUED

### ☐ RBC Enzymopathy Panel

(AK1, ALDOA, G6PD, GCLC, GPI, GPX1, GSR, GSS, HK1, NT5C3A, PFKM, PGK1, PKLR, TP11)

### ☐ 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](http://www.cincinnatichildrens.org/service/g/genetics-genomics-diagnostic-lab/requisition)

(ABCG5, ABCG8, ACTB, ACTN1, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S6, CDC42, CYCS, DIAPH1, DTNBP1, ETV6, FERMT3, FLI1, FLNA, FYB1, GALE, GATA1, GF11B, GNE, GPIBA, GPIBB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF5, ITGA2, ITGA2B, ITGB3, KDSR, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEA, NBEAL2, ORAI1, P2RX1, P2RY1, P2RY12, PLA2G4A, PRKACG, PTGS1, PTPRJ, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, STX11, STXBP2, TBXA2R, TBXAS1, THPO, TPM4, TUBB1, UNC13D, VIPAS39, VPS33B, VPS45, WAS)

### Thrombophilia Testing

#### ☐ Factor V (Leiden)

#### ☐ 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, CFHR2, CFHR3 and CFHR4 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, GF11B, HBA1, HBA2, HBB, HIF1A, HIF1AN (F1H), 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, BRIPI, 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, GF11, GLRX5, GPIBA, GPIBB, 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, NOPI0, NSMCE3, PALB2, PARN, PAX5, PGM3, POT1, PTPN11, PUS1, RAB27A, RAC2, RAD51, RAD51C, RBM8A, RFW3, 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, BRIPI, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, LIG4, MAD2L2, MYSM1, NBN, NHEJ1, NSMCE3, PALB2, RAD51, RAD51C, RFW3, 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, NOPI0, PARN, POT1, RTEL1, STN1, TERC, TERF2IP, TERT, TINF2, WRAP53)

### ☐ Immunology Exome

Gene sequencing panel with 475 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](http://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, RASGRP1, 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, BRIPI, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFW3, 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, 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 Gene Sequencing Panel

(AK2, AP3B1, CD40LG, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, EIF2AK3, ELANE, G6PC3, GATA1, GATA2, GF11, HAX1, HYOU1, JAGN1, LAMTOR2, LYST, MRTFA (MKL1), RAB27A, RAC2, RMRP, RUNX1, SBDS, SLC37A4, SMARCD2 (except exon 1), 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)

## 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, DGLUOK, 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](http://www.cincinnatichildrens.org/service/g/genetics-genomics-diagnostic-lab/requisition)
- ☐ Rubinstein-Taybi and Related Syndromes Gene Panel  
(*CREBBP, EP300, HNRNP1, HNRNP2, 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)
- ☐ 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

## 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*, *HCR1*, *HCRTR2*, *HDC*, *HLF*, *HOMER1*, *HOMER2*, *HTR1A*, *HTR1B*, *HTR2A*, *HTR2C*, *HTR7*, *HTT*, *IFNAR1*, *IL1R1*, *IL6*, *JAML*, *KANSL1*, *KCNA2*, *KCNA3*, *KCNK1*, *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*, *BMPRI1B*, *BPNT2*, *BRAF*, *BRD4*, *C2CD3*, *CBFB*, *CCNG1*, *CCNG2*, *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*, *HNRNP1K*, *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*, *PPPIR12A*, *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*, *SNRNP*, *SON*, *SOST*, *SOX11*, *SOX6*, *SOX9*, *SPECC1L*, *SPRY1*, *SPRY4*, *STAG2*, *STIL*, *SUFU*, *SUMO1*, *TBC1D32*, *TBX1*, *TBX22*, *TCF12*, *TCOF1*, *TFAP2A*, *TFAP2B*, *TGDS*, *TGFB1*, *TGFB2*, *TGFB3*, *TGFBF1*, *TGFBF2*, *TGFI1*, *TLK2*, *TMCO1*, *TOPORS*, *TP63*, *TRAF7*, *TRRAP*, *TWIST1*, *TWIST2*, *TXNL4A*, *UBE3B*, *USP9X*, *VAX1*, *VCAN*, *WASHC5*, *WDR19*, *WDR35*, *WNT5A*, *YAP1*, *YWHA*, *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](http://www.cincinnatichildrens.org/service/g/genetics-genomics-diagnostic-lab/requisition)  
(*ABCC6*, *ACTA2*, *ACVRL1*, *ADA2* (CECR1), *ATPIA2*, *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*, *P2RY2*, *PCNT*, *PDCD10*, *PMM2*, *POLG*, *PRRT2*, *PTPN11*, *RAF1*, *RASA1*, *RBBP8*, *RNF213*, *SAMHD1*, *SCN1A*, *SCN5A*, *SETD5*, *SLC19A2*, *SLC2A10*, *SMAD2*, *SMAD3*, *SMAD4*, *SMARCA1*, *SOS1*, *SUOX*, *TGFB2*, *TGFB3*, *TGFBF1*, *TGFBF2*, *TREX1*, *TSC1*, *TSC2*, *TTC19*, *WFS1*, *YYIAP1*)

- ☐ *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)