Cincinnati Children's
changing the outcome together

MOLECULAR GENETICS 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

Shipping Address:
3333 Burnet Avenue, Room R1042
Cincinnati, OH 45229

PRIMARY IMMUNODEFICIENCIES 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: CCHMC Patients, CCHMC 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

Note: For post-transplant patients, we accept pre-transplant samples or post-transplant skin fibroblasts **ONLY** (blood, saliva, and cytobrushes are not accepted). Culturing of skin fibroblasts is done at an additional charge.

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.

Cincinnati Children's
changing the outcome together

Patient Name: _____ Date of Birth: _____

INDICATIONS/DIAGNOSIS/ICD-9 CODE

Reason for Testing:

☐ Mutation detection in suspected affected patient

☐ Carrier testing

☐ Prenatal diagnosis (by previous arrangement only)

TEST(S) REQUESTED

Primary Immunodeficiency (Comprehensive testing)

☐ **Immunology Exome**

351 gene panel utilizing Whole Exome Sequencing (WES) technology*

☐ Reflex to deletion/duplication of single gene(s) (specify): _____

_____

*See page 5 for comprehensive gene list

Autoimmune lymphoproliferative syndrome

☐ Autoimmune Lymphoproliferative Syndrome (ALPS) Panel by next generation sequencing (NGS)

(CASP8, CASP10, FADD, FAS, FASLG, ITK, KRAS, MAGT1, NRAS)

☐ Reflex to deletion/duplication of all available genes on panel

☐ Reflex to deletion/duplication of single gene(s) (specify): _____

_____

☐ *FAS* (*TNFRSF6*)

☐ Reflex to deletion/duplication of *FAS (TNFRSF6)*

☐ *FASLG* (*TNFSF6*)

☐ Reflex to deletion/duplication of *FASLG (TNFSF6)*

☐ *CASP10*

☐ Reflex to deletion/duplication of *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

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

(AP3B1, BRCA2 (FANCD1), BRIP1 (FANCJ), CSF3R, CXCR4, DKC1, ELANE, ERCC4 (FANCQ), FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2 (ROBLD3), LYST, MPL, NHP2 (NOLA2), NOP10 (NOLA3), PALB2 (FANCN), RAB27A, RAC2, RAD51C (FANCO), RBM8A, RMRP, RPL5, RPL11, RPL15, RPL26, RPL35A, RPS7, RPS10, RPS17, RPS19, RPS24, RPS26, RTEL1, SBDS, SLC37A4, SLX4 (FANCP), SRP72, TAZ, TERC (hTR), TERT, TINF2, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53 (TCAB1, WDR79))

☐ Reflex to deletion/duplication of all available genes on panel†

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

_____

☐ Shwachman Diamond syndrome (*SBDS*)

☐ **Chromosome breakage syndrome panel**

(ATM, BLM, LIG4, NBN, NHEJ1)

☐ Reflex to deletion/duplication of all available genes on panel

☐ Reflex to deletion/duplication of single gene(s) (specify): _____

_____

☐ **Diamond-Blackfan anemia panel**

(GATA1, RPL5, RPL11, RPL15, RPL26, RPL35A, RPS7, RPS10, RPS17, RPS19, RPS24, RPS26)

☐ Reflex to deletion/duplication of all available genes on panel†

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

_____

†See page 5 for additional deletion/duplication information

☐ **Dyskeratosis congenita Panel**

(DKC1, NHP2 (NOLA2), NOP10 (NOLA3), RTEL1, TERC (hTR), TERT, TINF2, WRAP53 (WDR79, TCAB1))

☐ Reflex to deletion/duplication of all available genes on panel

☐ Reflex to deletion/duplication of single gene(s) (specify): _____

_____

Familial hemophagocytic lymphohistiocytosis

☐ Hemophagocytic Lymphohistiocytosis (HLH) Panel

(AP3B1, BLOC1S6, CD27, GATA2, ITK, LYST, MAGT1, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D (MUNC13-4), XIAP (BIRC4))

☐ Reflex to deletion/duplication of all available genes on panel

☐ Reflex to deletion/duplication of single gene(s) (specify): _____

_____

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*)

☐ Reflex to deletion/duplication of *UNC13D (MUNC13-4)*

___ *PRF1*

☐ Reflex to deletion/duplication of *PRF1*

___ *RAB27A* (Griscelli syndrome)

☐ Reflex to deletion/duplication of *RAB27A*

___ *STXBP2*

☐ Reflex to deletion/duplication of *STXBP2*

___ *STX11*

☐ Reflex to deletion/duplication of *STX11*

Fanconi anemia

☐ Fanconi Anemia Panel by next-generation sequencing (NGS)

(BRCA2 (FANCD1), BRIP1 (FANCJ), ERCC4 (FANCQ), FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2 (FANCN), RAD51C (FANCO), SLX4 (FANCP))

☐ Reflex to deletion/duplication of all available genes on panel†

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

_____

☐ *FANCA*

☐ Reflex to deletion/duplication of *FANCA*

☐ *FANCC*

☐ Reflex to deletion/duplication of *FANCC*

☐ *FANCC* c.456+4A>T (IVS4+4 A>T) [common Ashkenazi mutation] only

☐ *FANCG*

☐ Reflex to deletion/duplication of *FANCG*

Lymphoproliferative disorders (Including EBV-Related)

☐ *SH2D1A*

☐ Reflex to deletion/duplication of *SH2D1A*

☐ *XIAP (BIRC4)*

☐ Reflex to deletion/duplication of *XIAP (BIRC4)*

☐ *ITK*

☐ Reflex to deletion/duplication of *ITK*

☐ *MAGT1*

☐ Reflex to deletion/duplication of *MAGT1*

Cincinnati Children's
changing the outcome together

Patient Name: _____ Date of Birth: _____

TEST(S) REQUESTED, CONTINUED

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)
☐ Reflex to deletion/duplication of all available genes on panel†
☐ Reflex to deletion/duplication of single gene(s)† (specify): _____
_____

☐ X-linked severe combined immunodeficiency (*IL2RG*)
☐ Reflex to deletion/duplication of *IL2RG*

Severe congenital neutropenia

☐ Inherited neutropenia panel by next-generation sequencing (NGS)
(AP3B1, CSF3R, CXCR4, ELANE (ELA2), G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2 (ROBLD3), LYST, RAB27A, RAC2, SBDS, SLC37A4, TAZ, USB1, VPS13B, VPS45, WAS, WIPF1)
☐ Reflex to deletion/duplication of all available genes on panel†
☐ Reflex to deletion/duplication of single gene(s)† (specify): _____
_____

☐ *ELANE* (*ELA2*)
☐ Reflex to deletion/duplication of *ELANE* (*ELA2*)
☐ *HAX1*
☐ Reflex to deletion/duplication of *HAX1*
☐ *WAS* (males only)
☐ Reflex to deletion/duplication of *WAS*

†See page 5 for additional deletion/duplication information

Note: Single gene sequencing is available for all genes listed in the next-generation panels.

Other Primary Immunodeficiencies

☐ IPEX syndrome (*FOXP3*)
☐ Reflex to deletion/duplication of *FOXP3*
☐ Wiskott-Aldrich syndrome (*WAS*)
☐ Reflex to deletion/duplication of *WAS*
☐ X-linked hyper IgM immunodeficiency (*CD40LG*)
☐ Reflex to deletion/duplication of *CD40LG*

Rare Immunodeficiencies

☐ *CTLA4*
☐ Reflex to deletion/duplication of *CTLA4*
☐ *GATA2*
☐ Reflex to deletion/duplication of *GATA2*
☐ *LRBA*
☐ Reflex to deletion/duplication of *LRBA*
☐ *PIK3CD*
☐ Reflex to deletion/duplication of *PIK3CD*
☐ *STAT3*
☐ Reflex to deletion/duplication of *STAT3*

☐ Targeted (family specific) mutation analysis for _____ gene
If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

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.

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 mutation analysis

Proband's name: _____

Proband's DOB: _____

Proband's mutation: _____

Patient's relation to proband: _____

If testing was **not** performed at CCHMC, 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 mutation: _____

Patient's relation to proband: _____

If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

Cincinnati Children's
changing the outcome together

Patient Name: _____ Date of Birth: _____

IMMUNE DEFICIENCIES, AUTOIMMUNE DISORDERS AND BONE MARROW FAILURE SYNDROMES

Clinical History is Required for all NGS Panels

CLINICAL HISTORY

Has patient received a bone marrow transplant?

☐ Yes

☐ No

If yes, date of bone marrow transplant _____

Percent engraftment _____

General

☐ Acute liver failure

☐ Fever(s)

☐ Failure to thrive

☐ (Hepato)splenomegaly

☐ Lethargy

☐ Respiratory insufficiency/failure

☐ Sudden unexplained coma/death

☐ Other; specify_____

☐ Age at diagnosis _____

Head and Neck

☐ Abnormal CT/MRI of brain; specify_____

☐ Dysmorphic facies

☐ Enlarged lymph nodes

☐ Microcephaly

☐ Oral leukoplakia

☐ Small lymph nodes and/or tonsils

☐ Thymic hypoplasia

☐ Other; specify_____

Skin

☐ Alopecia

☐ Eczema

☐ Hypopigmentation/ hyperpigmentation

☐ Rash/dermatitis

☐ Telangiectasia of eyes or skin

☐ Dysplastic nails

☐ Other skin lesions; specify_____

Hematologic History

☐ Bone marrow failure

☐ Cytopenias (2 of 3 cell lineages)

☐ Leukopenia/neutropenia

☐ Red cell anemia

☐ Thrombocytopenia/small platelets

☐ Other; specify_____

Oncologic History

☐ Lymphoma; specify type _____

☐ Myelodysplasia/AML

☐ Other leukemia; specify type _____

☐ Recurrent primary tumors; specify types _____

☐ Solid tumor; specify type _____

☐ Other; specify _____

Infectious Disease History

☐ Recurrent, unusual or difficult to treat infections

___viral ___bacterial ___fungal

☐ Recurrent pneumonia, ear infections or sinusitis

☐ Recurrent deep abscesses of the organs or skin

☐ Multiple courses of antibiotics or IV antibiotics necessary to clear infections

☐ Other; specify_____

Laboratory findings

☐ Anemia

☐ Decreased telomere length

☐ Neutropenia/leukopenia

☐ Thrombocytopenia

☐ Abnormal ALPS panel

☐ Abnormal mitogen stimulation

☐ Abnormal lymphocyte subsets

☐ Abnormal TREC assay

☐ Abnormal B cell function; specify _____

☐ Abnormal T cell function; specify _____

☐ Low or absent NK function

☐ Complementation group correction (specify) _____

☐ Increased chromosome breakage

☐ ↑ ferritin

☐ ↑ soluble IL2Rα

☐ ↑ triglycerides and/or ↓fibrinogens

☐ Abnormal protein assay by flow cytometry; specify _____

☐ Other; specify_____

Congenital abnormalities/malformations/dysmorphic features

(Please specify)

_____

_____

_____

_____

Other Symptoms *(Please specify)*

_____

_____

_____

_____

Related disease history of other family members *(Please specify)*

_____

_____

_____

_____

Cincinnati Children's
changing the outcome together

ADDITIONAL INFORMATION

IMMUNOLOGY EXOME PANEL — GENES TESTED

ACP5	*ACTB*	*ADA*	*ADA2*	*ADAMTS13*	*ADAR*	*AICDA*	*AIRE*	*AK2*	*AP3B1*
APOL1	*ATM*	*BLM*	*BLNK*	*BLOC1S3*	*BLOC1S6*	*BRCA2*	*BRIP1*	*BTK*	*C1QA*
C1QB	*C1QC*	*C1S*	*C2*	*C3*	*C4BPA*	*C5*	*C6*	*C7*	*C8A*
C8B	*C8G*	*C9*	*CARD11*	*CARD14*	*CARD9*	*CARMIL2*	*CASP10*	*CASP8*	*CAVIN1*
CD19	*CD247*	*CD27*	*CD3D*	*CD3E*	*CD3G*	*CD40*	*CD40LG*	*CD46*	*CD59*
CD70	*CD79A*	*CD79B*	*CD81*	*CD8A*	*CDCA7*	*CEBPE*	*CFB*	*CFD*	*CFH*
CFHR1	*CFHR3*	*CFHR5*	*CFI*	*CFP*	*CHD7*	*CIITA*	*CLEC7A*	*CLPB*	*COG6*
COLEC11	*CORO1A*	*CR2*	*CREBBP*	*CSF3R*	*CTC1*	*CTLA4*	*CTPS1*	*CTSC*	*CXCR4*
CYBA	*CYBB*	*DCLRE1C*	*DGKE*	*DHFR*	*DKC1*	*DNAJC21*	*DNMT3B*	*DOCK8*	*DTNBP1*
ELANE	*EPG5*	*ERCC2*	*ERCC3*	*ERCC4*	*ERCC6L2*	*ETV6*	*F11*	*F13A1*	*F13B*
F5	*F7*	*F8*	*F9*	*FADD*	*FANCA*	*FANCB*	*FANCC*	*FANCD2*	*FANCE*
FANCF	*FANCG*	*FANCI*	*FANCL*	*FANCM*	*FAS*	*FASLG*	*FCN3*	*FERMT3*	*FGA*
FGB	*FOXN1*	*FOXP3*	*FPR1*	*G6PC*	*G6PC3*	*G6PD*	*GATA1*	*GATA2*	*GFI1*
GP1BA	*GP1BB*	*GP9*	*GTF2H5*	*HAX1*	*HELLS*	*HPS1*	*HPS3*	*HPS4*	*HPS5*
HPS6	*ICOS*	*IFIH1*	*IFNGR1*	*IFNGR2*	*IGLL1*	*IKBKB*	*IKZF1*	*IL10*	*IL10RA*
IL10RB	*IL12B*	*IL12RB1*	*IL17F*	*IL17RA*	*IL1RN*	*IL2*	*IL21R*	*IL2RA*	*IL2RG*
IL36RN	*IL7R*	*INSR*	*IRAK4*	*IRF8*	*ISG15*	*ITCH*	*ITGAM*	*ITGB2*	*ITK*
JAGN1	*JAK2*	*JAK3*	*KMT2D*	*KRAS*	*LAMTOR2*	*LCK*	*LIG1*	*LIG4*	*LPIN2*
LRBA	*LRRC8A*	*LYST*	*MAGT1*	*MALT1*	*MAN2B1*	*MANBA*	*MASP1*	*MASP2*	*MBL2*
MC2R	*MCM4*	*MEFV*	*MLPH*	*MPL*	*MPO*	*MRE11*	*MS4A1*	*MTHFD1*	*MVK*
MYD88	*MYH9*	*MYO5A*	*MYSM1*	*NBN*	*NCF2*	*NCF4*	*NCSTN*	*NFKB1*	*NFKB2*
NFKBIA	*NHEJ1*	*NHP2*	*NKX2-5*	*NLRC4*	*NLRP12*	*NLRP3*	*NOD2*	*NOP10*	*NRAS*
ORAI1	*PALB2*	*PARN*	*PCCA*	*PCCB*	*PEPD*	*PGM3*	*PI4KA*	*PIGA*	*PIK3CD*
PIK3R1	*PLCG2*	*PLG*	*PMM2*	*PNP*	*POLE*	*PRF1*	*PRKCD*	*PRKDC*	*PROC*
PROS1	*PSENEN*	*PSMB8*	*PSTPIP1*	*PTPRC*	*RAB27A*	*RAC2*	*RAD50*	*RAD51C*	*RAG1*
RAG2	*RBCK1*	*RBM8A*	*RFX5*	*RFXANK*	*RFXAP*	*RHOH*	*RNASEH2A*	*RNASEH2B*	*RNASEH2C*
RNF168	*RPL11*	*RPL15*	*RPL26*	*RPL35A*	*RPL36*	*RPL5*	*RPS10*	*RPS15*	*RPS15A*
RPS17	*RPS19*	*RPS24*	*RPS26*	*RPS27A*	*RPS28*	*RPS29*	*RPS7*	*RPSA*	*RTEL1*
RUNX1	*SAMHD1*	*SBDS*	*SEMA3E*	*SH2D1A*	*SH3BP2*	*SKIV2L*	*SLC29A3*	*SLC35A1*	*SLC35C1*
SLC37A4	*SLC39A4*	*SLC46A1*	*SLC7A7*	*SLX4*	*SMARCAL1*	*SP110*	*SPINK5*	*SRP72*	*STAT1*
STAT2	*STAT3*	*STAT5B*	*STIM1*	*STK4*	*STX11*	*STXBP2*	*TAP1*	*TAP2*	*TAPBP*
TAZ	*TBK1*	*TBX1*	*TCIRG1*	*TCN2*	*TERC*	*TERT*	*THBD*	*TICAM1*	*TINF2*
TLR3	*TMC6*	*TMC8*	*TNFAIP3*	*TNFRSF13B*	*TNFRSF13C*	*TNFRSF1A*	*TNFRSF4*	*TNFSF12*	*TRADD*
TRAF3	*TRAF3IP2*	*TREX1*	*TRNT1*	*TTC37*	*TTC7A*	*TYK2*	*UNC119*	*UNC13D*	*UNG*
USB1	*VPS13B*	*VPS45*	*WAS*	*WDR1*	*WIPF1*	*WRAP53*	*XIAP*	*XK*	*ZAP70*
ZBTB24									

†Deletion/Duplication analysis is currently unavailable for the following genes: *ACP5, ACTB, ADA, ADAR, APOL1, C8G, CARMIL2, CAVIN1, CD46, CD70, CFH, CFHR1, CFHR3, CFHR5, CLPB, CORO1A, CTC1, CTPS1, DCLRE1C, DHFR, DNAJC21, EPG5, ERCC6L2, ETV6, FANCD2, FERMT3, GTF2H5, HELLS, IFIH1, IGLL1, IKBKB, IL2, ITGAM, JAGN1, KMT2D, MC2R, MRE11, MYSM1, NCSTN, NFKB1, NFKB2, NLRC4, PARN, PEPD, PGM3, PI4KA, PIGA, POLE, PRKCD, PROS1, PSENEN, RPL15, RPL36, RPS15, RPS15A, RPS17, RPS28, RPS29, RPSA, RUNX1, SBDS, SEMA3E, SKIV2L, SLC29A3, SLC39A4, STAT2, STAT5B, TNFRSF4, TNFSF12, TRADD, TRAF3IP2, TRNT1, WDR1.*