

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.

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 (FANCI), CSF3R, CXCR4, DKC1, ELANE, ERCC4 (FANCG), 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): _____

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 (FANCI), ERCC4 (FANCG), 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*

*See page 5 for additional deletion/duplication information

TEST(S) REQUESTED, CONTINUED

Severe Combined Immunodeficiencies

- Severe combined immunodeficiency panel by next-generation sequencing (NGS)
(ADA, CD3D, CD3E, DCLRE1C, FOXP3, 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.

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

Complement group correction (specify) _____

Increased chromosome breakage

↑ ferritin

↑ soluble IL2R α

↑ 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)*

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	GF11
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	PSENE1	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, PSENE1, RPL15, RPL36, RPS15, RPS15A, RPS17, RPS28, RPS29, RPSA, RUNX1, SBDS, SEMA3E, SKIV2L, SLC29A3, SLC39A4, STAT2, STAT5B, TNFRSF4, TNFSF12, TRADD, TRAF3IP2, TRNT1, WDR1.