Cincinnati Children's

MOLECULAR GENETICS LABORATORY

For courier service and/or inquiries, please contact 513-636-4474 • Fax: 513-636-4373
www.cincinnatichildrens.org/moleculargenetics • Email: moleculargenetics@cchmc.org

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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)

☐ PATIENT BILLING

☐ Check Enclosed ☐ Money Order ☐ Credit Card (Visa, MC, Amex., Disc.)

Credit Card Number: _____

Card Holder Name: _____

Expiration Date: _____

Signature: _____

☐ REFERRING INSTITUTION

Institution: _____

Address: _____

City/State/Zip: _____

Accounts Payable Contact Name: _____

Phone: _____

Fax: _____

Email: _____

☐ COMMERCIAL INSURANCE/ POLICY HOLDER INFORMATION*

Name: _____

Gender: _____ Date of Birth _____ / _____ / _____

Authorization Number: _____

Insurance Name: _____

Insurance Address: _____

City/State/Zip: _____

Insurance ID Number: _____

Group Number: _____

Insurance Phone Number: _____

* PLEASE NOTE:

- We will bill Medicaid or Medicaid HMO only for tests ordered for Cincinnati Children's patients by Cincinnati Children's providers.
- Insurance can only be billed if requested at the time of service
- Please call 866-450-4198 with any billing questions.

SAMPLE/SPECIMEN INFORMATION

Has patient received a bone marrow transplant? ☐ Yes ☐ No

If yes, date of bone marrow transplant _____

Percent engraftment _____

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.

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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

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 entire 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 and 513-636-4685 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, WRAP53 (TCAB1, WDR79))
- ☐ Reflex to deletion/duplication of entire 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 entire 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 entire 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 entire panel
- ☐ Reflex to deletion/duplication of single gene(s) (specify): _____

_____

Familial hemophagocytic lymphohistiocytosis

☐ Hemophagocytic Lymphohistiocytosis (HLH) Panel
(AP3B1, BLOC1S6, CD27, ITK, LYST, MAGT1, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D (MUNC13-4), XIAP (BIRC4))
- ☐ Reflex to deletion/duplication of entire 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 entire 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* 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*

†Deletion/Duplication analysis of *DCLRE1C, FANCD2, RPL15, RPS17,* and *SBDS* is not available at this time.

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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 entire 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 entire 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*

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*

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

☐ **Targeted (family specific) mutation analysis of genes listed above**
Gene of interest _____
Proband's name _____
Proband's DOB _____
Proband's mutation _____
Relationship to proband _____

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

†Deletion/Duplication analysis of *DCLRE1C, FANCD2, RPL15, RPS17,* and *SBDS* is not available at this time.

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
☐ Targeted analysis for a common mutation seen in the population of interest
Mutation: _____
☐ Familial mutation analysis
Proband's name: _____
Proband's DOB: _____
Proband's mutation: _____
Patient's relation to proband: _____
Please include proband's report, if not performed at CCHMC.

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
☐ Targeted deletion or duplication analysis for a common mutation seen in the population of interest
Mutation: _____
☐ 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: _____
Please include proband's report, if not performed at CCHMC.

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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_____

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)*

_____

_____

_____

_____