Complete and print.

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MAYO CLINIC LABORATORIES

Primary Immunodeficiencies Patient Information

Instructions: The accurate interpretation and reporting of genetic results is contingent upon the reason for referral, clinical information, ethnic background, and family history. To help provide the best possible service, supply the information requested below and **send paperwork with the specimen, or return by fax to Mayo Clinic Laboratories, Attn: Personalized Genomics Laboratory Genetic Counselors at 507-284-1759. Phone: 507-266-5700 / International clients: +1-507-266-5700 or email mclglobal@mayo.edu**

Patient Information

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*	Sex ☐ Male ☐ Female
Referring Provider Name *(Last, First)*	Phone	Fax*
Other Contact Name *(Last, First)*	Phone	Fax*

**Fax number given must be from a fax machine that complies with applicable HIPAA regulations.*

Reason for Testing

☐ Diagnosis ☐ Newborn Screening Follow-up ☐ Carrier Testing ☐ Family History

Note: Genetic testing should always be initiated on an affected family member first, when available, in order to be most informative for at-risk relatives.

Indications Check all that apply.

Autoinflammatory

- ☐ Periodic fever
- ☐ Familial Mediterranean fever (FMF)
- ☐ Hyper IgD syndrome
- ☐ Cryopyrin-associated periodic syndromes (CAPS)
- ☐ Blau syndrome
- ☐ PAPA syndrome
- ☐ PFAPA syndrome
- ☐ TRAPS (TNF-receptor-associated periodic syndromes)
- ☐ PLAID/APLAID
- ☐ Amylopectinosis and autoinflammation
- ☐ Majeed syndrome; CRMO
- ☐ Other inflammasome-related disorders
- ☐ Other autoinflammatory conditions, specify: _____

B-Cell Deficiency; Agammaglobulinemia

- ☐ Recurrent sinopulmonary infections
- ☐ Hypogammaglobulinemia
- ☐ Lymphoproliferation
- ☐ Increased IgM (Hyper IgM)
- ☐ Class-switch recombination defects

Complement aHUS/TMA

- ☐ Atypical hemolytic uremic syndrome (aHUS)
- ☐ Thrombotic microangiopathy (TMA)
- ☐ Thrombotic thrombocytopenic purpura (TTP)

Inflammatory Bowel Disease/ Enteropathy/Hepatic PID

- ☐ Chronic IBD-like disorder and CID
- ☐ Ulcerative colitis
- ☐ Crohn disease
- ☐ Enteropathy, hypogammaglobulinemia, autoinflammation, and autoimmunity
- ☐ IBD, lymphadenopathy
- ☐ Veno-occlusive disease (in context of PID; VODI)
- ☐ NRH (nodular regenerative hyperplasia)

Phagocytic PID/ Chronic Granulomatous Disease

- ☐ Recurrent pneumonia, soft-tissue granulomas, recurrent abscesses, specific microbial infections; specify: _____
- ☐ Palmoplantar keratoderma with periodontitis (Papillon-Lefvre)
- ☐ Delayed umbilical cord separation +/- omphalitis
- ☐ Leukocytosis
- ☐ Absence of pus (leukocyte adhesion deficiencies)
- ☐ Bleeding diathesis
- ☐ Comel-Netherton syndrome
- ☐ Favism (hemolysis, neonatal hyperbilirubinemia)
- ☐ Pulmonary alveolar proteinosis
- ☐ Other neutrophil-associated phenotypes
- ☐ Bombay blood group
- ☐ Gingivitis
- ☐ Periodontitis

Severe Combined Immunodeficiency

- ☐ Severe combined immunodeficiency (SCID)
- ☐ Combined immunodeficiency (CID)
- ☐ T-cell lymphopenia/deficiency
- ☐ (T-, B-, NK-) SCID (ADA-SCID; Reticular dysgenesis)
- ☐ (T-, B-, NK+) SCID (VDJ recombination defects; CID)
- ☐ (T-, B+, NK-) SCID (X-linked SCID; JAK3 SCID)
- ☐ (T-, B+, NK+) SCID (T-cell SCID)
- ☐ Severe, recurrent EBV infections/ ENB lymphoproliferative disease
- ☐ CD4+ or CD8+ T-cell deficiency or absence of MHC class I or class II molecules (Bare lymphocyte syndrome, type I or II)

Telomere Defects

- ☐ Idiopathic pulmonary fibrosis
- ☐ Dyskeratosis congenita
- ☐ Bone marrow failure syndrome
- ☐ Telomeropathies

Congenital Neutropenia/Neutrophil PID

- ☐ Congenital neutropenia (Kostmann syndrome)
- ☐ Cyclic neutropenia
- ☐ Shwachman-Diamond syndrome
- ☐ Wiskott-Aldrich syndrome
- ☐ Cohen syndrome
- ☐ Barth syndrome
- ☐ G6PD deficiency
- ☐ WHIM syndrome
- ☐ Other neutropenia, specify: _____

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Primary Immunodeficiencies Patient Information (continued)

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Family History Attach Pedigree if available.

Are there any affected relatives? ☐ Yes ☐ No
If yes, relationship: _____

Is there any consanguinity in the family? ☐ Yes ☐ No
Have relatives had molecular genetic testing? ☐ Yes ☐ No
If yes, specify: _____

Ethnicity

☐ European Caucasian ☐ African American ☐ Hispanic ☐ Asian ☐ Other: _____

Clinical History Check all that apply.

Age of onset of symptoms: _____ Durations of symptoms: _____

Has the patient received a hematopoietic cell transplant? ☐ Yes ☐ No If yes, transplant date *(mm-dd-yyyy)*: _____

Transplant type: allogeneic (MRD, MURD, haplo, cord, BM): _____

Donor chimerism: % T: _____ % B: _____ % NK: _____ % myeloid: _____

Has the patient received a solid organ transplant? ☐ Yes ☐ No
If yes, ☐ Heart ☐ Lung ☐ Liver ☐ Kidney ☐ Vascularized Composite allograph
☐ Other, specify: _____

First transplant or multiple: _____

Post-transplant immunosuppression
Graft versus host disease? ☐ Yes: ☐ Acute ☐ Chronic ☐ No

Laboratory Findings

☐ Abnormal lymphocyte (T-, B-, and NK-cell) subset quantitation:
_____

Humoral Markers

☐ Abnormal B-cell function (vaccine antibody responses):
_____

☐ Autoantibodies present, specify:
_____

☐ Hypogammaglobulinemia:
☐ IgG ☐ IgA ☐ IgM
☐ IgD ☐ IgE

☐ Hypergammaglobulinemia:
☐ IgG ☐ IgA ☐ IgM
☐ IgD ☐ IgE

Cellular Markers

☐ Abnormal TREC assay (NBS and/or other): _____

☐ Abnormal T-cell function (specify mitogens/antigens/anti-CD3/cytokine production);

☐ T-cell markers:
Naive: ☐ Increased ☐ Decreased
Memory: ☐ Increased ☐ Decreased
Activated:
☐ Increased ☐ Decreased

☐ B-cell markers:
Switched memory:
☐ Increased ☐ Decreased
Marginal zone B-cells:
☐ Increased ☐ Decreased
Transitional B-cells:
☐ Increased ☐ Decreased
Plasmablasts:
☐ Increased ☐ Decreased

Cytokines

IL-1b: ☐ Increased ☐ Decreased
IL-6: ☐ Increased ☐ Decreased
IL-18: ☐ Increased ☐ Decreased
TNF alpha: ☐ Increased ☐ Decreased
Interferon-gamma:
☐ Increased ☐ Decreased

Chromosomal Studies

☐ 22q deletion FISH
☐ Chromosomal array
☐ Other chromosomal abnormality
_____

Protein Loss Markers

☐ Calprotectin
☐ 24-hour stool alpha-1 antitrypsin clearance assay
☐ Serum albumin
☐ Proteinuria: ☐ Yes ☐ No

Soluble Biomarkers

☐ ADAMTS13
Activity: _____ Level: _____
☐ Shiga toxin: ☐ Positive ☐ Negative
☐ Vitamin B12: _____
☐ Folate: _____
☐ Ferritin: _____
☐ Soluble IL2R-alpha (sCD25): _____
☐ CRP: _____
☐ ESR: _____
☐ Triglycerides: _____
☐ Fibrinogen: _____
☐ AFP level (age when tested): _____
☐ ALPS screening panel:
DNT-cell % as % CD3+ : _____
sFASL: ☐ Increased ☐ Abnormal

Complement Serology

☐ CH50: ☐ Normal ☐ Abnormal
☐ AH50: ☐ Normal ☐ Abnormal
☐ FH autoantibody: ☐ Yes ☐ No
☐ FH: ☐ Normal ☐ Abnormal
☐ FB: ☐ Normal ☐ Abnormal
☐ FI: ☐ Normal ☐ Abnormal
☐ FD: ☐ Normal ☐ Abnormal
☐ sMAC: ☐ Normal ☐ Abnormal
☐ aHUS serology panel
C2 level: _____
Function: ☐ Normal ☐ Abnormal
C3 level: _____
Function: ☐ Normal ☐ Abnormal
C4 level: _____
Function: ☐ Normal ☐ Abnormal
C5 level: _____
Function: ☐ Normal ☐ Abnormal
C6–C9 level: _____
Function: ☐ Normal ☐ Abnormal
C1q level: _____
Function: ☐ Normal ☐ Abnormal
C1q antibody: ☐ Yes ☐ No
C3NeF: ☐ Yes ☐ No
Other: _____

Other Markers

☐ Abnormal radiosensitivity: ☐ Yes ☐ No (blood, MB, or fibroblasts)
☐ Specific protein assay by flow cytometry:
BTK: ☐ Normal ☐ Abnormal
LRBA: ☐ Normal ☐ Abnormal
DOCK8: ☐ Normal ☐ Abnormal
WAS: ☐ Normal ☐ Abnormal
XIAP: ☐ Normal ☐ Abnormal
SAP: ☐ Normal ☐ Abnormal

Primary Immunodeficiencies Patient Information (continued)

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Other Markers (continued)

- ☐ Functional studies:
  - T-cell: ☐ Normal ☐ Abnormal (specify mitogens/antigens/anti-CD3/ cytokine production)
  - STAT signaling: ☐ Normal ☐ Abnormal
  - DHR: ☐ Normal ☐ Abnormal
  - Calcium signaling: ☐ Normal ☐ Abnormal
  - Other: _____
- ☐ Telomere length:
  - Lymphoid: ☐ Normal ☐ <10% ☐ <1%
  - Myeloid: ☐ Normal ☐ <10% ☐ <1%
- ☐ ADA1:
  - Activity: _____ Level: _____
- ☐ ADA2:
  - Activity: _____ Level: _____
- ☐ PNP:
  - Activity: _____ Level: _____

Other findings:

Infectious Disease History

- ☐ Recurrent, 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 necessary to clear infections
- ☐ On replacement immunoglobulin

Hematologic/Vascular History

- ☐ Lymphopenia: subset and count (cells/mcL): _____
- ☐ Lymphocytosis: subset and count (cells/mcL): _____
- ☐ Leukocytosis: _____
- ☐ Bone marrow failure
- ☐ Bone marrow biopsy
  - Abnormalities seen: ☐ Yes ☐ No
- ☐ Marrow cellularity: _____
- ☐ Myelokathexis
- ☐ Agranulocytosis
- ☐ Neutrophilia
- ☐ Cytopenias (2 of 3 cell lineages)
- ☐ Leukopenia/neutropenia
- ☐ Thrombocytopenia/small platelets
- ☐ Anemia
- ☐ Lymphedema
- ☐ Aneurysms
- ☐ Polyarteritis nodosa (PAN)
- ☐ Stroke
- ☐ Anti-platelet antibody
- ☐ Anti-granulocyte antibody
- ☐ COOMBS test: ☐ Positive ☐ Negative
- ☐ Lymphangiectasia, location: _____

Oncologic History

- ☐ Myelodysplasia/AML
- ☐ Lymphoma, specify: _____
- ☐ Solid tumor, specify: _____
- ☐ Leukemia, specify: _____
- ☐ Recurrent primary tumors, specify: _____
- ☐ Other: _____

Head and Neck

- ☐ Dysmorphic facies, specify: _____
- ☐ Lymphadenopathy
- ☐ Microcephaly
- ☐ Oral leukoplakia
- ☐ Small lymph nodes and/or tonsils
- ☐ Thymic hypoplasia
- ☐ Abnormal CT/MRI of brain: _____
- ☐ Other: _____

Skin/Hair

- ☐ Albinism
- ☐ Alopecia
- ☐ Dysplastic nails
- ☐ Ectodermal dysplasia
- ☐ Eczema
- ☐ Hypopigmentation/hyperpigmentation
- ☐ Rash/dermatitis
- ☐ Telangiectasia of eyes or skin
- ☐ Other: _____

General History

- ☐ Acute liver failure
- ☐ Cardiac surgery in infancy or early childhood
- ☐ Dental anomalies
- ☐ Failure to thrive
- ☐ Fever(s)
- ☐ Fontan procedure
- ☐ (Hepato) splenomegaly
- ☐ Lethargy
- ☐ Protein losing enteropathy
- ☐ Respiratory insufficiency/failure
- ☐ Skeletal anomalies
- ☐ Sudden unexplained coma/death
- ☐ Thymectomy

Other Clinical Symptoms:

Medications

On immunosuppressant therapy?
☐ Yes ☐ No

Previous immunosuppressant therapy:
Duration: _____
Date of use: _____

Other Comments:

Attach clinical notes if available.

Print