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MAYO CLINIC LABORATORIES

Combined Immunodeficiency, Severe Combined Immunodeficiency, and B-Cell/Antibody Deficiency Patient Information

Instructions: Accurate interpretation and reporting of genetic results is contingent upon the reason for testing, clinical information, family history, and ancestry. 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: Molecular Technologies Laboratory Genetic Counselors at 507-284-1759. Phone: 800-533-1710 / International clients: +1-507-266-5700 or email MLIINT@mayo.edu**

Patient Information

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

Sex Assigned at Birth	Legal/Administrative Sex
☐ Male ☐ Female ☐ Unknown ☐ Choose not to disclose	☐ Male ☐ Female ☐ Nonbinary

Referring Provider Information

Referring Provider Name (Last, First)	Phone	Fax*
Genetic Counselor Name (Last, First)	Phone	Fax*

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

Reason for Testing Specify below or attach relevant clinic note.

☐ Confirm clinical diagnosis, specify diagnosis: Age of onset:

☐ Newborn screening follow-up

☐ Family history**, describe:

☐ Other, specify:

**Genetic testing should be performed on an affected family member first, when available. FMTT / Familial Mutation Targeted Testing should be ordered when there is a previous positive genetic test result in the family.

Infectious Disease History

☐ Recurrent or difficult to treat infections: ☐ Viral ☐ Bacterial ☐ Fungal

☐ Recurrent pneumonia, ear infections, sinusitis or other sinopulmonary infections

☐ Recurrent deep abscesses of the organs or skin

☐ Gastrointestinal infections

☐ Skin infections, describe:

☐ Conjunctivitis

☐ Meningitis

☐ Sepsis

☐ Other infection, specify:

☐ Multiple courses of antibiotics necessary to clear infection

☐ On immunoglobulin replacement

Combined Immunodeficiency, Severe Combined Immunodeficiency, and B-Cell/Antibody Deficiency Patient Information (continued)

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

Laboratory Findings

☐ Abnormal lymphocyte (T-, B-, and NK-cell) subset quantitation; describe or attach report:

Humoral markers:

☐ Abnormal B-cell function (vaccine antibody responses)

☐ Autoantibodies present, specify:

☐ Immunoglobulins:

☐ IgG: ☐ Increased ☐ Decreased

☐ IgG1: ☐ Increased ☐ Decreased

☐ IgG2: ☐ Increased ☐ Decreased

☐ IgG3: ☐ Increased ☐ Decreased

☐ IgG4: ☐ Increased ☐ Decreased

☐ IgA: ☐ Increased ☐ Decreased

☐ IgM: ☐ Increased ☐ Decreased

☐ IgD: ☐ Increased ☐ Decreased

☐ IgE: ☐ Increased ☐ Decreased

Cellular markers:

☐ Abnormal TREC assay (eg, newborn screening)

☐ Abnormal T-cell function: ☐ Mitogens ☐ Antigens ☐ Anti-CD3 ☐ Anti-CD3/CD28 ☐ Cytokine production

☐ T-cell subsets:

☐ Naive: ☐ Increased ☐ Decreased

☐ Memory: ☐ Increased ☐ Decreased

☐ Activated: ☐ Increased ☐ Decreased

☐ B-cell subsets:

☐ Naive: ☐ Increased ☐ Decreased

☐ Memory: ☐ Increased ☐ Decreased

☐ Switched memory: ☐ Increased ☐ Decreased

☐ Marginal zone B-cells: ☐ Increased ☐ Decreased

☐ Transitional B-cells: ☐ Increased ☐ Decreased

☐ Plasmablasts: ☐ Increased ☐ Decreased

☐ Oligoclonal T-cells or abnormal TCRVB spectratyping

☐ Abnormal CD4 T-cell recent thymic emigrants, flow cytometry

☐ Abnormal haemophilus influenzae B vaccine response

☐ Abnormal HLA typing for class I or class II HLA antigens

☐ Abnormal streptococcus pneumoniae IgG antibody response

Specific protein assay by flow cytometry:

☐ BTK: ☐ Normal ☐ Abnormal

☐ LRBA: ☐ Normal ☐ Abnormal

☐ DOCK8: ☐ Normal ☐ Abnormal

☐ WAS: ☐ Normal ☐ Abnormal

☐ XIAP: ☐ Normal ☐ Abnormal

☐ SAP: ☐ Normal ☐ Abnormal

☐ Other, specify:

Blood:

☐ Autoimmune cytopenia ☐ Eosinophilia ☐ Lymphocytosis ☐ Lymphopenia ☐ Thrombocytopenia

☐ Other hematological abnormality, specify:

☐ Other laboratory findings, specify:

Combined Immunodeficiency, Severe Combined Immunodeficiency, and B-Cell/Antibody Deficiency Patient Information (continued)

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

Oncologic History

- ☐ Myelodysplasia/AML
- ☐ Lymphoma, specify:
- ☐ Solid tumor, specify:
- ☐ Leukemia, specify:
- ☐ Skin cancer, specify:
- ☐ Other, specify:
- ☐ Family history of cancer; specify cancer type and biological relationship to patient:

General History

- ☐ Alopecia
- ☐ Ataxia
- ☐ Bone abnormalities, describe:
- ☐ Bronchiectasis
- ☐ Celiac disease
- ☐ Decreased lymphoid tissue (small adenoids, tonsils, lymph nodes)
- ☐ Diarrhea
- ☐ Eczema
- ☐ Endodrine abnormalities, describe:
- ☐ Enteropathy, describe:
- ☐ Erythroderma
- ☐ Failure to thrive
- ☐ Graft vs host disease
- ☐ Granulomas
- ☐ Hepatosplenomegaly
- ☐ Lymphadenopathy
- ☐ Oral candidiasis
- ☐ Osteopenia
- ☐ Pruritic dermatitis
- ☐ Telangiectasia
- ☐ Thymic defect, describe:

Patient Treatment History

Has the patient received an allogenic stem cell transplant***? ☐ No ☐ Yes; transplant date (mm-dd-yyyy):

Is the patient transfusion-dependent***? ☐ No ☐ Yes; last transfusion date (mm-dd-yyyy):
Was this transfusion leukoreduced***? ☐ No ☐ Yes ☐ Unknown

Chemotherapy: ☐ No ☐ Yes; date (mm-dd-yyyy):

***Results may be inaccurate due to the presence of donor DNA if the patient has received an allogeneic hematopoietic stem cell transplant or non-leukocyte reduced blood products. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Family History

Are there similarly affected relatives? ☐ Yes ☐ No
If "Yes," indicate relationship, and diagnosis or symptoms:

Have any family member had genetic testing? ☐ Yes**** ☐ No ☐ Unknown
****FMTT / Familial Mutation Targeted Testing should be ordered when there is a previous positive genetic test result in the family. Contact the lab for ordering assistance.**

History of consanguinity: ☐ No ☐ Yes; relationship details:

Ancestry

- ☐ African/African American
- ☐ East Asian
- ☐ Latinx/Latine
- ☐ South Asian
- ☐ Unknown
- ☐ Ashkenazi Jewish
- ☐ European
- ☐ Middle Eastern
- ☐ None of the above
- ☐ Choose not to disclose

New York State Patients: Informed Consent for Genetic Testing is required. See Informed Consent for Genetic Testing (T576), Informed Consent for Genetic Testing – Spanish (T826), or Informed Consent for Genetic Testing for Deceased Individuals (T782).

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