

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: 855-379-3115 or +1-507-284-9273, or email mliintl@mayo.edu.**

Patient Information

Patient Name (Last, First Middle)		Birth Date (mm-dd-yyyy)
Sex Assigned at Birth <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Unknown <input type="checkbox"/> Choose not to disclose		Legal/Administrative Sex <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Nonbinary

Referring Healthcare Professional Information

Referring Healthcare Professional 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.

<input type="checkbox"/> Confirm clinical diagnosis, specify diagnosis: _____ Age of onset: _____ <input type="checkbox"/> Newborn screening follow-up <input type="checkbox"/> Family history**, describe: _____ <input type="checkbox"/> Other, specify: _____
<p>**Genetic testing should be performed on an affected family member first, when available. FMTT / Familial Variant, Targeted Testing should be ordered when there is a previous positive genetic test result in the family.</p>

Infectious Disease History

<input type="checkbox"/> Recurrent or difficult to treat infections: <input type="checkbox"/> Viral <input type="checkbox"/> Bacterial <input type="checkbox"/> Fungal <input type="checkbox"/> Recurrent pneumonia, ear infections, sinusitis or other sinopulmonary infections <input type="checkbox"/> Recurrent deep abscesses of the organs or skin <input type="checkbox"/> Gastrointestinal infections <input type="checkbox"/> Skin infections, describe: _____ <input type="checkbox"/> Conjunctivitis <input type="checkbox"/> Meningitis <input type="checkbox"/> Sepsis <input type="checkbox"/> Other infection, specify: _____ <input type="checkbox"/> Multiple courses of antibiotics necessary to clear infection <input type="checkbox"/> On immunoglobulin replacement

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Patient Information (continued)

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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 IgG3: Increased Decreased
 IgG2: 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 Activated: Increased Decreased
 Memory: Increased Decreased

B-cell subsets:

Naive: Increased Decreased Marginal zone B-cells: Increased Decreased
 Memory: Increased Decreased Transitional B-cells: Increased Decreased
 Switched memory: 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 WAS: Normal Abnormal
 LRBA: Normal Abnormal XIAP: Normal Abnormal
 DOCK8: Normal Abnormal SAP: Normal Abnormal

Other, specify: _____

Blood:

Autoimmune cytopenia Eosinophilia Lymphocytosis Lymphopenia Thrombocytopenia

Other hematological abnormality, specify: _____

Other laboratory findings, specify: _____

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Oncologic History

<input type="checkbox"/> Myelodysplasia/AML	<input type="checkbox"/> Leukemia, specify: _____
<input type="checkbox"/> Lymphoma, specify: _____	<input type="checkbox"/> Skin cancer, specify: _____
<input type="checkbox"/> Solid tumor, specify: _____	<input type="checkbox"/> Other, specify: _____
<input type="checkbox"/> Family history of cancer; specify cancer type and biological relationship to patient: _____	

General History

<input type="checkbox"/> Alopecia	<input type="checkbox"/> Failure to thrive
<input type="checkbox"/> Ataxia	<input type="checkbox"/> Graft vs host disease
<input type="checkbox"/> Bone abnormalities, describe: _____	<input type="checkbox"/> Granulomas
<input type="checkbox"/> Bronchiectasis	<input type="checkbox"/> Hepatosplenomegaly
<input type="checkbox"/> Celiac disease	<input type="checkbox"/> Lymphadenopathy
<input type="checkbox"/> Decreased lymphoid tissue (small adenoids, tonsils, lymph nodes)	<input type="checkbox"/> Oral candidiasis
<input type="checkbox"/> Diarrhea	<input type="checkbox"/> Osteopenia
<input type="checkbox"/> Eczema	<input type="checkbox"/> Pruritic dermatitis
<input type="checkbox"/> Endocrine abnormalities, describe: _____	<input type="checkbox"/> Telangiectasia
<input type="checkbox"/> Enteropathy, describe: _____	<input type="checkbox"/> Thymic defect, describe: _____
<input type="checkbox"/> Erythroderma	

Patient Treatment History

Has the patient received an allogeneic stem cell transplant***? <input type="checkbox"/> No <input type="checkbox"/> Yes; transplant date (mm-dd-yyyy): _____
Is the patient transfusion-dependent****? <input type="checkbox"/> No <input type="checkbox"/> Yes; last transfusion date (mm-dd-yyyy): _____ Was this transfusion leukoreduced***? <input type="checkbox"/> No <input type="checkbox"/> Yes <input type="checkbox"/> Unknown
Chemotherapy: <input type="checkbox"/> No <input type="checkbox"/> 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? <input type="checkbox"/> Yes <input type="checkbox"/> No If "Yes," indicate relationship, and diagnosis or symptoms: _____
Have any family member had genetic testing? <input type="checkbox"/> Yes**** <input type="checkbox"/> No <input type="checkbox"/> Unknown ****FMTT / Familial Variant, 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: <input type="checkbox"/> No <input type="checkbox"/> Yes; relationship details: _____

Ancestry

<input type="checkbox"/> African/African American	<input type="checkbox"/> East Asian	<input type="checkbox"/> Latinx/Latine	<input type="checkbox"/> South Asian	<input type="checkbox"/> Unknown
<input type="checkbox"/> Ashkenazi Jewish	<input type="checkbox"/> European	<input type="checkbox"/> Middle Eastern	<input type="checkbox"/> None of the above	<input type="checkbox"/> 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).