



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

Form with fields for Patient Name (Last, First Middle), Birth Date (mm-dd-yyyy), Sex Assigned at Birth (Male, Female, Unknown, Choose not to disclose), and Legal/Administrative Sex (Male, Female, Nonbinary).

Referring Provider Information

Form with fields for Referring Provider Name (Last, First), Phone, Fax*, Genetic Counselor Name (Last, First), Phone, and 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.

Form with checkboxes for Confirm clinical diagnosis, Family history, and Other, specify. Includes a note: **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.

Clinical Findings

Form with checkboxes for Crohn's Disease, Inflammatory Bowel Disease - Unclassified, Ulcerative Colitis, and other symptoms/disorders. Includes fields for Age of onset and descriptions for Malabsorption, Endocrine abnormalities, Failure to thrive, Fever, Hepatosplenomegaly, Skin, hair, dental, or nail findings, Uveitis, and Venous-occlusive disease.

Infectious Disease History

Form with checkboxes for Recurrent or difficult to treat infections (Viral, Bacterial, Fungal), Recurrent deep abscesses of the organs or skin, Gastrointestinal infections, Other infection, specify, and On immunoglobulin replacement.

Early Onset Inflammatory Bowel Disease

Patient Information (continued)

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

Fecal Calprotectin: Normal Increased

IgA class anti gliadin antibodies: Present Absent

IgA class antitransglutaminase antibodies (tTGA): Present Absent

IgA class endomysial antibodies: Present Absent

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: _____

If the patient has had GI biopsies, attach a copy of the pathology report.

Early Onset Inflammatory Bowel Disease

Patient Information (continued)

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

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).