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MAYO CLINIC LABORATORIES

# *Inborn Errors of Immunity, Autoimmunity, and Autoinflammatory Disease 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 800-533-1710. Phone: 507-266-5700 / 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, or sinusitis ☐ Multiple courses of antibiotics necessary to clear infections
☐ Recurrent deep abscesses of the organs or skin ☐ On immunoglobulin replacement

## Laboratory Findings

☐ Abnormal TREC assay (eg, newborn screening)
☐ Abnormal lymphocyte (T-, B-, and NK-cell) subset quantitation:
☐ Autoimmune lymphoproliferative syndrome (ALPS) workup:
- ☐ Alpha/Beta TCR positive CD4 CD8 Double Negative T-cells % of CD3+:
- ☐ sFasL > 200 pg/mL
- ☐ IL-10 > 20 pg/mL
- ☐ IL-18 > 500 pg/mL
- ☐ Vitamin B12 > 1500 ng/L

☐ T-cell immunophenotyping:
☐ Abnormal T-cell function: ☐ Mitogens ☐ Antigens ☐ Anti-CD3 ☐ Cytokine production
☐ Abnormal DHR
☐ Immunoglobulins:

| | | | | | | |
|---|---|---|---|---|---|---|
| ☐ IgG: | ☐ Increased | ☐ Decreased | ☐ IgD: | ☐ Increased | ☐ Decreased |
| ☐ IgA: | ☐ Increased | ☐ Decreased | ☐ IgE: | ☐ Increased | ☐ Decreased |
| ☐ IgM: | ☐ Increased | ☐ Decreased | | | |

Blood:
- ☐ Leukocytosis
- ☐ Monoclonal lymphocytosis
- ☐ Lymphopenia
- ☐ Neutropenia (Neutrophils < $1 \times 10^9$/L): ☐ Cyclic ☐ Persistent ☐ Congenital ☐ Acquired
- ☐ Neutrophilia
- ☐ Pancytopenia
- ☐ Thrombocytopenia (Platelets < $100 \times 10^9$/L): ☐ Congenital ☐ Acquired
- ☐ Hemolytic anemia
- ☐ Sideroblastic anemia
- ☐ Other hematological abnormality; specify:

☐ Other laboratory findings; specify:

# *Inborn Errors of Immunity, Autoimmunity, and Autoinflammatory Disease Patient Information* (continued)

## General History

| | |
|---|---|
| ☐ Alopecia | ☐ Inflammatory bowel disease |
| ☐ Atopy (allergies); specify: | ☐ Intellectual disability |
| ☐ Candidiasis | ☐ Joint pain |
| ☐ Conjuctivitis | ☐ Liver failure |
| ☐ Dental anomalies | ☐ Lung disease, specify: |
| ☐ Dysmorphic facies | ☐ Lymphadenopathy |
| ☐ Eczema | ☐ Lymphoproliferation |
| ☐ Encephalitis | ☐ Meningitis |
| ☐ Failure to thrive | ☐ Osteopetrosis |
| ☐ Fever; duration: frequency: | ☐ Panniculitis |
| triggers: | ☐ Polyendocrinopathy |
| ☐ Folliculitis | ☐ Skeletal anomalies, specify: |
| ☐ Growth failure | ☐ Solid organ autoimmunity |
| ☐ Hepatitis | ☐ Systemic lupus erythematosus |
| ☐ Hyperextensible joints | ☐ Type 1 Diabetes |
| ☐ Inflammatory arthritis | ☐ Other; specify: |

## Oncologic History

| | |
|---|---|
| ☐ Myelodysplasia/AML | ☐ Leukemia; specify: |
| ☐ Lymphoma; specify: | ☐ Skin cancer; specify: |
| ☐ Solid tumor; specify: | ☐ Other; specify: |

## 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 members 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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