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Reset Form

MAYO CLINIC LABORATORIES

Congenital Neutropenia, Bone Marrow Failure, Telomere Defects, and Pulmonary Fibrosis (IPF) 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:

☐ Family history**; describe:

☐ Other; specify:

**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.

Infectious Disease History

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

☐ Recurrent pneumonia, ear infections, or sinusitis

☐ Recurrent deep abscesses of the organs or skin

Laboratory Findings

☐ Bone marrow biopsy: ☐ Normal ☐ Abnormal; describe or attach report:

☐ T-cell immunophenotyping:

☐ Telomere length studies; method: ☐ Flow FISH ☐ Other; specify:

- ☐ Lymphoid: ☐ Normal ☐ < 10% ☐ < 1%
- ☐ Myeloid: ☐ Normal ☐ < 10% ☐ < 1%

☐ Increased chromosomal breakage of peripheral blood lymphocytes in the presence of DNA cross-linking agents, such as mitomycin C or diepoxybutane

☐ Immunoglobulins:

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

Blood:

- ☐ Abnormally elevated fetal hemoglobin (Hb F) for age
- ☐ Erythrocytosis
- ☐ Macrocytic anemia
- ☐ Megaloblastic anemia
- ☐ Normocytic anemia
- ☐ Sideroblastic anemia
- ☐ Neutropenia: ☐ Cyclic ☐ Persistent ☐ Congenital ☐ Acquired
  ☐ Mild (1 to 1.5 × 10^9/L) ☐ Moderate (0.5 to 1 × 10^9/L) ☐ Severe (< 0.5 × 10^9/L)
- ☐ Lymphopenia
- ☐ Thrombocytopenia (platelets < 100 × 10^9/L): ☐ Congenital ☐ Acquired
- ☐ Macrothrombocytopenia
- ☐ Small-platelet thrombocytopenia
- ☐ Pancytopenia
- ☐ Other hematological abnormality; specify:

☐ Other laboratory findings; specify:

Congenital Neutropenia, Bone Marrow Failure, Telomere Defects, and Pulmonary Fibrosis (IPF) Patient Information (continued)

Oncologic History

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

General History

- ☐ Abnormal skin pigmentation; describe:
- ☐ Aplastic anemia
- ☐ Bilateral exudative retinopathy
- ☐ Cardiomyopathy or heart defect; describe:
- ☐ Cellulitis
- ☐ Cerebellar hypoplasia
- ☐ Chronic hypersensitivity pneumonitis
- ☐ Cirrhosis
- ☐ Developmental delay
- ☐ Dysmorphic facies
- ☐ Dysplastic nails
- ☐ Eczema
- ☐ Exocrine pancreatic dysfunction
- ☐ Gastrointestinal disease; specify:
- ☐ Gingivitis
- ☐ Hemophagocytic lymphohistiocytosis (HLH)
- ☐ Hypogammaglobulinemia
- ☐ Iron overload
- ☐ Liver disease
- ☐ Neonatal respiratory distress
- ☐ Neurological dysfunction; describe:
- ☐ Omphalitis
- ☐ Oral leukoplakia
- ☐ Oral ulcers
- ☐ Osteomyelitis
- ☐ Premature graying hair
- ☐ Pulmonary hypertension
- ☐ Pulmonary fibrosis
- ☐ Recurrent fevers
- ☐ Red cell aplasia
- ☐ Reticular dysgenesis
- ☐ Short stature
- ☐ Skeletal abnormalities; describe:
- ☐ Thymic hypoplasia
- ☐ Urogenital abnormalities; describe:
- ☐ Vasculopathy
- ☐ Warts
- ☐ 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): _____

Note: Skin biopsy is the preferred specimen type to detect germline variants in patients with active hematological malignancy.

***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 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: ☐ 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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