



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

Instructions: This form is intended to be completed by the ordering healthcare professional. Accurate interpretation and reporting of genetic results is contingent upon the reason for testing, clinical information, biological 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)
Preferred Name	Medical Record Number (if Birth Date is not available)	
Sex Assigned at Birth ☐ Male ☐ Female ☐ Choose not to disclose ☐ Other, specify: _____	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary ☐ Choose not to disclose ☐ Other, specify: _____	
Gender Identity (optional)	Pronouns (optional)	

Referring Healthcare Professional Information

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

Reason for Testing Specify below or attach relevant clinic note.

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

☐ Confirm clinical diagnosis; specify diagnosis: _____	Age of onset: _____
☐ Biological family history**, describe: _____	
☐ Other; specify: _____	

**Genetic testing should be performed on an affected biological 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
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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 ($1 \text{ to } 1.5 \times 10^9/\text{L}$) ☐ Moderate ($0.5 \text{ to } 1 \times 10^9/\text{L}$) ☐ Severe ($< 0.5 \times 10^9/\text{L}$) ☐ Lymphopenia ☐ Thrombocytopenia (platelets $< 100 \times 10^9/\text{L}$): ☐ Congenital ☐ Acquired ☐ Macrothrombocytopenia ☐ Small-platelet thrombocytopenia ☐ Pancytopenia ☐ Other hematological differences; specify: _____
☐ Other laboratory findings; specify: _____

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Patient Information (continued)

Oncologic History

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

General History

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

Biological Family History

Are there similarly affected biological relatives? ☐ Yes ☐ No	
If "Yes," indicate relationship, and diagnosis or symptoms: _____	
Have any biological 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 biological family.	
Contact the lab for ordering assistance.	
History of consanguinity: ☐ No ☐ Unknown ☐ Choose not to disclose ☐ Yes; relationship details: _____	

Patient Ancestry

Ancestry: _____	☐ Unknown ☐ Choose not to disclose
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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).