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**Reporting Title:** Congenital Neutropenia GenePanel**Performing Location:** Rochester**Ordering Guidance:**

Targeted testing for familial variants (also called site-specific or known variants testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about testing option, call 800-533-1710.

**Shipping Instructions:**

Specimen preferred to arrive within 96 hours of collection.

**Specimen Requirements:**

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblasts

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

**Specimen Minimum Volume:**

Blood: 1 mL; Skin biopsy or cultured fibroblasts: See Specimen Required

**Forms:**

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
  - Informed Consent for Genetic Testing (T576)
  - Informed Consent for Genetic Testing (Spanish) (T826)
2. Molecular Genetics: Congenital Inherited Diseases Patient Information (T521)
3. Congenital Neutropenia, Bone Marrow Failure, Telomere Defects, and Pulmonary Fibrosis (IPF) Patient Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

**Ask at Order Entry (AOE) Questions:**

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
619873	Test Description	Alphanumeric		62364-5
619874	Specimen	Alphanumeric		31208-2
619875	Source	Alphanumeric		31208-2
619876	Result Summary	Alphanumeric		50397-9
619877	Result	Alphanumeric		82939-0
619878	Interpretation	Alphanumeric		69047-9
619879	Additional Results	Alphanumeric		82939-0
619880	Resources	Alphanumeric		99622-3
619881	Additional Information	Alphanumeric		48767-8
619882	Method	Alphanumeric		85069-3

Result ID	Reporting Name	Type	Unit	LOINC®
619883	Genes Analyzed	Alphanumeric		82939-0
619884	Disclaimer	Alphanumeric		62364-5
619885	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

## Supplemental Report:

Supplemental

## CPT Code Information:

81443  
88233- Tissue culture, skin, solid tissue biopsy (if appropriate)  
88240- Cryopreservation (if appropriate)

## Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes

## Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

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**Reference Values:**

An interpretive report will be provided.