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**Reporting Title:** Telomere Disorders Gene Panel**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, Chorionic Villi/Products of Conception/Tissue. 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, Chorionic Villi/Products of Conception/Tissue. 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®
619887	Test Description	Alphanumeric		62364-5
619888	Specimen	Alphanumeric		31208-2
619889	Source	Alphanumeric		31208-2
619890	Result Summary	Alphanumeric		50397-9
619891	Result	Alphanumeric		82939-0
619892	Interpretation	Alphanumeric		69047-9
619893	Additional Results	Alphanumeric		82939-0
619894	Resources	Alphanumeric		99622-3

Result ID	Reporting Name	Type	Unit	LOINC®
619895	Additional Information	Alphanumeric		48767-8
619896	Method	Alphanumeric		85069-3
619897	Genes Analyzed	Alphanumeric		82939-0
619898	Disclaimer	Alphanumeric		62364-5
619899	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

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Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

**Reference Values:**

An interpretive report will be provided