
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.

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 | | |

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 |
| 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 | Available Separately |
|---------|-------------------------------------|-----------|----------|------------------|----------------------|
| CULFB | Fibroblast Culture for Genetic Test | 1 | 88233 | 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 |

Reference Values:

An interpretive report will be provided