
Reporting Title: SMN1 Full Gene Analysis**Performing Location:** Rochester**Ordering Guidance:**

This is **not** the preferred genetic test for carrier screening or diagnosis in individuals with suspicion of spinal muscular atrophy (SMA). For these situations, order SMNCS / Spinal Muscular Atrophy Carrier Screening, Deletion/Duplication Analysis, Varies or SMNDX / Spinal Muscular Atrophy Diagnostic Assay, Deletion/Duplication Analysis, Varies.

This test is appropriate for second-tier carrier screening following SMNCS / Spinal Muscular Atrophy Carrier Screening, Deletion/Duplication Analysis, Varies when:

- There is a family history of SMA, but an affected individual is not available for testing
- The disease-causing variants are unknown
- Testing the reproductive partner of a known SMA carrier

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)/Refrigerated

Specimen Type: Cultured fibroblasts

Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 Full T-75 or 2 full T-25 flasks

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.

Supplies: Fibroblast Biopsy Transport Media (T115)

Specimen Type: Skin biopsy

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.

- Supplies:** Card - Blood Spot Collection (Filter Paper) (T493)
- Specimen Type:** Blood spot
- Container/Tube:**
- Preferred:** Collection card (Whatman Protein Saver 903 Paper)
- Acceptable:** PerkinElmer 226 (formerly Ahlstrom 226) filter paper or blood spot collection card
- Specimen Volume:** 5 Blood spots
- Collection Instructions:**

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

- Additional Information:**
1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.
 2. For collection instructions, see [Blood Spot Collection Instructions](#)
 3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)
 4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)

- 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 in Special Instructions:
[-Informed Consent for Genetic Testing](#) (T576)
[-Informed Consent for Genetic Testing-Spanish](#) (T826)
 2. [Molecular Genetics: Congenital Inherited Diseases Patient Information](#) (T521) in Special Instructions.

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Varies | Varies | | |

Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|-----------|------------------------|--------------|------|---------|
| 602754 | Result Summary | Alphanumeric | | 50397-9 |
| 602755 | Result | Alphanumeric | | 82939-0 |
| 602756 | Interpretation | Alphanumeric | | 69047-9 |
| 602757 | Additional Information | Alphanumeric | | 48767-8 |
| 602758 | Specimen | Alphanumeric | | 31208-2 |
| 602759 | Source | Alphanumeric | | 31208-2 |
| 602760 | Released By | Alphanumeric | | 18771-6 |

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81336
88233-Tissue culture, skin, or 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.