

---

**Reporting Title:** Mitochondrial Deletion Heteroplasmy  
**Performing Location:** Rochester

**Ordering Guidance:**  
For diagnosis of a mitochondrial DNA deletion syndrome, the recommended first tier test is MITOP/ Mitochondrial Full Genome Analysis, Next-Generation Sequencing (NGS), Varies.

**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:** Cultured fibroblasts  
**Container/Tube:** T-25 flask  
**Specimen Volume:** 2 Flasks  
**Collection Instructions:** Submit confluent cultured fibroblast cells from a skin biopsy. 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 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:** Muscle tissue biopsy

**Supplies:** Muscle Biopsy Kit (T541)

**Collection Instructions:** Prepare and transport specimen per instructions in [Muscle Biopsy Specimen Preparation Instructions](#).

**Specimen Volume:** 10-80 mg

**Specimen Stability Information:** Frozen (preferred)/Ambient/Refrigerated

**Specimen Type:** Snap frozen nerve tissue biopsy

**Collection Instructions:** Prepare snap frozen tissue biopsy per surgical procedure

**Specimen Volume:** 0.25-0.5 cm

**Specimen Stability Information:** Frozen

**Specimen Type:** Blood spot

**Supplies:** Card-Blood Spot Collection (Filter Paper) (T493)

**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:** 2 to 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:  
[-Informed Consent for Genetic Testing](#) (T576)  
[-Informed Consent for Genetic Testing \(Spanish\)](#) (T826)

2. [Molecular Genetics: Biochemical Disorders Patient Information](#) (T527)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
618613	Result Summary	Alphanumeric		50397-9
618614	Result	Alphanumeric		82939-0
618615	Interpretation	Alphanumeric		69047-9
618616	Additional Information	Alphanumeric		48767-8
618617	Specimen	Alphanumeric		31208-2
618618	Source	Alphanumeric		31208-2
618619	Method	Alphanumeric		85069-3
618620	Disclaimer	Alphanumeric		62364-5
618621	Released By	Alphanumeric		18771-6

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

Supplemental Report:

No

CPT Code Information:

81479

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	No

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.