
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).

Specimen Minimum Volume:

Blood: 1 mL; Blood spots: 2 spots; Other specimen types: 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: Biochemical Disorders Patient Information (T527)

Specimen Type	Temperature	Time	Special Container
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Varies	Varies	
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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®
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	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			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.