MAYO CLINIC LABORATORIES

TEST CHANGE

Notification Date: December 3, 2024
Effective Date: January 2, 2025

Mitochondrial Full Genome Analysis, Next-Generation Sequencing (NGS), Varies

Test ID: MITOP

Explanation: On the effective date, this assay will incorporate changes to the Specimen Required, as shown below. Of note, the Whole Blood specimen type will no longer accept *any anticoagulant* and will be limited to the anticoagulants shown below. Ordering guidance will also be updated.

Current Specimen Required	New Specimen Required
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** **Preferred:** Lavender top (EDTA) tube or yellow top (ACD) tube **Acceptable:** Any anticoagulant **Specimen Volume:** 3 mL **Collection Instructions:** 1. Invert several times to mix blood. 2. Send 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. **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	**Patient Preparation:** A previous bone marrow transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a bone marrow transplant, call 800-533-1710. **Submit only 1 of the following specimens:** **Specimen Type: Whole blood** **Preferred:** Lavender top (EDTA) or yellow top (ACD) **Acceptable:** Green Top (Sodium heparin) **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 4 days/Refrigerated 4 days/ Frozen 4 days **Additional Information:** **1.** Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed. 2. To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate. **Specimen Type**: **Cultured fibroblasts** **Source:** Skin **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) <24 hours/Refrigerated <24 hours **Additional Information**: 1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for

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)
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)

specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are 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: Ambient (preferred) <24 hours/Refrigerated <24 hours
Additional Information:
1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts before genetic testing can occur.

Specimen Type: **Snap frozen nerve tissue biopsy**
Collection Instructions: Prepare snap frozen tissue biopsy per surgical procedure
Specimen Volume: 0.25 to 0.5 cm
Specimen Stability Information: Frozen

Specimen Type: **Muscle tissue biopsy**
Supplies: Muscle Biopsy Kit (T541)
Specimen Volume: 20 to 80 mg
Collection Instructions: Prepare and transport specimen per instructions in Muscle Biopsy Specimen Preparation.
Specimen Stability Information: Frozen (preferred) <24 hours/Ambient <24 hours/Refrigerated <24 hours
Additional Information: Specimens are preferred to be received within 24 hours of collection. Extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.

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 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 a Dried Blood Spot Sample.
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. Blood spot specimens are acceptable but not recommended. Multiple extractions will be required to obtain sufficient yield for supplemental analysis, and there is significant risk for test failure due to insufficient DNA. 2. Due to lower concentration of DNA yielded from blood spot, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing. 3. For collection instructions, see Blood Spot Collection Instructions 4. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777) 5. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800) **Specimen Type:** Extracted DNA **Container/Tube:** **Preferred:** Screw Cap Micro Tube, 2 mL with skirted conical base **Acceptable:** Matrix tube, 1 mL **Collection Instructions:** 1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL. 2. Include concentration and volume on tube. Specimen Stability Information: Frozen (preferred) 1 year/Ambient/Refrigerated **Additional Information:** DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Current Ordering Guidance	New Ordering Guidance
If testing for variants in the mitochondrial genes encoded by the nuclear genome is requested, order MITON / Mitochondrial Nuclear Gene Panel, Next-Generation Sequencing (NGS), Varies	If testing for variants in the mitochondrial genes encoded by the nuclear genome is requested, order NMITO / Nuclear Mitochondrial Gene Panel, Next-Generation Sequencing, Varies. Alternatively, order CMITO / Combined Mitochondrial Full Genome and Nuclear Gene Panel, Varies for both the mitochondrial genome and mitochondrial genes encoded by the nuclear genome.

Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.