MAYO CLINIC LABORATORIES

TEST CHANGE

Notification Date: November 19, 2024
Effective Date: December 24, 2024

Friedreich Ataxia, Repeat Expansion Analysis, Varies

Test ID: AFXN

Explanation: On the effective date, this assay will incorporate changes to the Specimen Required, as shown below. The Whole Blood specimen type will no longer accept *any anticoagulant* and will be limited to the anticoagulants shown below. Blood Spot specimen type will have an update to the Specimen Volume and will no longer be located in the Prenatal Specimens section. In addition, all Specimen Types will include updates to the Stability and/or Additional Information fields. The days performed 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. For instructions for 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) 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.** **Prenatal Specimens** **Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.** **Specimen Type: Amniotic fluid** **Container/Tube:** Amniotic fluid container **Specimen Volume:** 20 mL **Specimen Stability Information:** Refrigerated (preferred)/Ambient **Additional information:** 1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid. **2. All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.	**Patient Preparation:** A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for 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:** None **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 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: Blood spot** **Supplies:** Card-Blood Spot Collection (Filter Paper) (T493) **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

Specimen Type: **Chorionic villi**
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated
Additional Information:
1. 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.
2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Acceptable:
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 Flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated
Additional Information: All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Blood spot
Supplies: Card-Blood Spot Collection (Filter Paper) (T493)
Container/Tube:
Preferred: Collection card (Whatman Protein Saver 903 Paper)
Acceptable: Perkin/Elmer 266 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. For collection instructions, see Blood Spot Collection Instructions
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

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)

PRENATAL SPECIMENS
Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
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/or 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 CULAF / Culture for Genetic Testing, Amniotic Fluid. An additional 2 to 3 weeks is required to culture amniotic fluid before genetic testing can occur.
3. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Ambient (preferred) <24 hours/Refrigerated <24 hours
Additional Information:

4. Due to lower concentration of DNA yielded from blood spots, it is possible that additional specimen may be required to complete testing.	1. Specimens are preferred to be received within 24 hours of collection. Culture and/or 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 is required to culture fibroblasts before genetic testing can occur. 3. **All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen. **Specimen Type: Cultured chorionic villi** **Container/Tube:** T-25 flasks **Specimen Volume:** 2 Full flasks **Collection Instructions:** Submit confluent cultured cells from another laboratory. **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/or 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. 3. **All prenatal specimens must be accompanied by a maternal blood specimen**; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Current Days Performed	New Days Performed
Monday, Wednesday	Monday

Questions

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