MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: February 22, 2022
Effective Date: March 15, 2022

Friedreich Ataxia, Repeat Expansion Analysis, Varies

Test ID: AFXN

Useful for:
Molecular confirmation of clinically suspected Friedreich ataxia

Reflex Tests:

Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No

Methods:
Polymerase Chain Reaction (PCR)

Reference Values:
FXN
Normal alleles: <34 GAA repeats
Borderline alleles: 34-65 GAA repeats
Expanded alleles: >65 GAA repeats

An interpretive report will be provided.

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
Minimum Volume:	0.5 mL

Specimen Type	**Amniotic fluid**
Container/Tube:	Amniotic fluid container
Specimen Volume:	20 mL
Specimen Stability Information:	Ambient (preferred)/Refrigerated
Minimum Volume:	10 mL

Specimen Type	**Chorionic villi**
Container/Tube:	15-mL tube containing 15 mL of transport media
Specimen Volume:	20 mg
Specimen Stability Information:	Refrigerated
Minimum Volume:	5 mg

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

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 (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 of age is a fingerstick. See Dried Blood Spot Collection Tutorial for how to collect blood spots via fingerstick: https://vimeo.com/508490782 . 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
Minimum Volume:	5 punches, 3-mm diameter
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). 4. Due to lower concentration of DNA yielded from blood spots, it is possible that additional specimen may be required to complete testing.

Note:

Due to its complexity, prenatal testing will not be accepted without approval from the laboratory. Call 800-533-1710 for prenatal testing consultation. Prenatal specimens can be sent Monday through Thursday and **must be received by 5 p.m. CST on Friday** in order to be processed appropriately. All prenatal specimens must be accompanied by a maternal blood specimen. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Cautions:

For familial testing, it is important to first document the molecular etiology of disease in an affected family member to confirm that a repeat expansion is the underlying mechanism of disease in the family. Specifically, this assay will not detect nonrepeat expansion variants (eg, sequence variants, deletions, and duplications).

It is strongly recommended that patients undergoing genetic testing receive genetic counseling.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data, such as frataxin concentrations (see FFRWB / Friedreich Ataxia, Frataxin, Quantitative, Blood and FFRBS / Friedreich Ataxia, Frataxin, Quantitative, Blood Spot). Errors in test interpretation may occur if the provided information is inaccurate or incomplete.

Rare variants (ie, polymorphisms) may exist, such as intron 1 deletions, which could lead to false-negative results. If GAA-repeat expansion results do not match clinical findings, additional testing should be considered.

Due to somatic mosaicism, GAA repeat-sizes in peripheral blood specimens may not reflect GAA repeat-sizes in other tissues (eg, central nervous system).

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories at 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

CPT Code:

81284
81265-Maternal Cell Contamination (if appropriate)

88233- Fibroblast Culture (if appropriate)
88235-Amniotic Fluid Culture (if appropriate)
88240-Cryopreservation (if appropriate)

Day(s) Performed: Monday & Wednesday **Report Available:** 8 to 16 days

Note:
The following referral test code(s) will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
Friedreich Ataxia Repeat Expansion Analysis - Unknown Mutation	FFRED	6031	Baylor Medical Genetics Laboratories

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