

Overview

Useful For

Diagnosing individuals with Friedreich ataxia in whole blood specimens

Monitoring frataxin levels in patients with Friedreich ataxia

Highlights

Friedreich ataxia (FA) presents most commonly between 10 to 15 years of age with progressive neurologic changes including spasticity and ataxia.

Decreased frataxin protein levels are diagnostic of FA and can also be utilized for monitoring known patients.

Fratxin protein analysis is a cost-effective and quick method for establishing a diagnosis of FA and will detect rare variants otherwise missed by common molecular-based trinucleotide repeat analysis.

Special Instructions

- [Informed Consent for Genetic Testing](#)
- [Biochemical Genetics Patient Information](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)

Method Name

LuminexImmunoassay

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Necessary Information

Provide a reason for referral with each specimen.

Specimen Required

Collection Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Green top (sodium or lithium heparin)

Submission Container/Tube:Plastic vial

Specimen Volume:2 mL

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 in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Biochemical Genetics Patient Information (T602) in Special Instructions.

3. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:

-[Neurology Specialty Testing Client Test Request](#) (T732)

-[Inborn Errors of Metabolism Test Request](#) (T798)

Specimen Minimum Volume

1.25 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time
Whole blood	Frozen (preferred)	70 days
	Ambient	70 days
	Refrigerated	70 days

Clinical and Interpretive

Clinical Information

Friedreich ataxia (FA) is an autosomal recessive disease affecting approximately 1:50,000 Caucasians. The disease is clinically characterized by progressive spasticity, ataxia, dysarthria, absent lower limb reflexes, sensory loss, and scoliosis. Hypertrophic cardiomyopathy is present in approximately two-thirds of patients and is the most frequent cause of premature death in individuals with FA. Although most individuals begin experiencing initial symptoms between 10 and 15 years of age, atypical late-onset forms with initial symptoms presenting after age 25 do occur.

FA is caused by mutations in the *FXN* gene encoding a mitochondrial protein, frataxin. Mutations in this gene lead to a reduced expression of frataxin, which causes the clinical manifestations of the disease. Approximately 98% of individuals with FA have a homozygous expansion of the GAA trinucleotide repeat in intron 1 of *FXN*. The remaining 2% of FA patients have the trinucleotide expansion on 1 allele and a point mutation or deletion on the second allele. Normal alleles contain between 5 to 33 GAA repeats. Disease-causing alleles typically range from 66 to 1,700 repeats, though the majority of individuals with FA have repeats ranging from 600 to 1,200.

Historically, FA has been diagnosed by use of a DNA-based molecular test to detect the presence of the GAA expansion. Unfortunately, testing for the triplet repeat expansion will miss those patients with point mutations or deletions. Moreover, a molecular-based analysis is not able to effectively monitor treatment, is not amenable to

multiplexing with other disease analytes, nor can it be efficiently utilized for population screening. In contrast, a protein-based assay measuring concentration of frataxin is suitable for both diagnosis as well as treatment monitoring in individuals with FA.

Reference Values

Pediatric (<18 years) normal frataxin: > or =19 ng/mL

Adults (> or =18 years) normal frataxin: > or =21 ng/mL

Interpretation

Normal results (> or =19 ng/mL for pediatric and > or =21 ng/mL for adult patients) in properly submitted specimens are not consistent with Friedreich ataxia.

For results outside the normal reference range an interpretative comment will be provided.

Cautions

This test is not suitable for carrier detection.

Clinical Reference

1. Deutsch EC, Oglesbee D, Greeley NR, Lynch DR: Usefulness of frataxin immunoassays for the diagnosis of Friedreich ataxia. *J Neurol Neurosurg Psychiatry* 2014 Sep;85(9):994-1002
2. Babady NE, Carelle N, Wells RD, et al: Advancements in the pathophysiology of Friedreich ataxia and new prospects for treatments. *Mol Genet Metab* 2007;92:23-35
3. Boehm T, Scheiber-Mojdehkar B, Kluge B, et al: Variations of frataxin protein levels in normal individuals. *Neurol Sci* 2011 Apr;32(2):327-330 doi: 10.1007/s10072-010-0326-1

Performance**Method Description**

The immunoassay utilizes frataxin-specific monoclonal antibodies bound to Luminex microspheres as capture antibodies and biotinylated frataxin-specific polyclonal antibodies as detection antibodies. Streptavidin-phycoerythrin attaches to the biotin and when exposed to light at 352 nM emits a photon that is measured and that signal is used to determine the amount of frataxin in the sample.(Deutsch EC, et al: A rapid, noninvasive immunoassay for frataxin: utility in assessment of Friedreich ataxia. *Mol Genet Metab* 2010, doi:10.1016/j.ymgme.2010.07.001)

PDF Report

No

Day(s) and Time(s) Test Performed

Alternating Fridays

Analytic Time

14 days

Maximum Laboratory Time

30 days

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

Fees and Codes**Fees**

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact [Customer Service](#).

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

83520

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
FFRWB	Frataxin, Quant, WB	80979-8

Result ID	Test Result Name	Result LOINC Value
32253	Reason for Referral	42349-1
32254	Method	49549-9
32255	Frataxin	80979-8
32256	Interpretation	59462-2