Overview

Useful For
Diagnosing individuals with Friedreich ataxia in whole blood specimens

Monitoring frataxin levels in patients with Friedreich ataxia

This test is not useful for carrier detection.

Genetics Test Information
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.

Highlights
Frataxin protein analysis is a cost-effective and quick method for establishing a diagnosis of Friedreich Ataxia (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
Luminex Immunoassay

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**

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<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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**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. Cardiac involvement occurs with the development of myocardial fibrosis due to mitochondrial proliferation and loss of contractile proteins. It tends to be correlated with the clinical neurologic age of onset and the GAA triplet repeat length, but not the duration of disease or the severity of neurologic symptoms. 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 1200.

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. 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**

No significant cautionary statements

**Clinical Reference**


**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;101(2-3):238-245 doi:10.1016/j.ymgme.2010.07.001)

**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Alternating Fridays
**Test Definition: FFRWB**

Frataxin, Quant, WB

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**Analytic Time**
14 days

**Maximum Laboratory Time**
30 days

**Specimen Retention Time**
1 month

**Performing Laboratory Location**
Rochester

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**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**

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