TEST ID: FFRBS

FRIEDREICH ATAXIA, FRATAXIN, QUANTITATIVE, BLOOD SPOT

USEFUL FOR

- Diagnosing individuals with Friedreich ataxia
- Monitoring frataxin levels in patients with Friedreich ataxia

CLINICAL INFORMATION

Friedreich ataxia (FA) is an autosomal recessive disease affecting approximately 1 in 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 with FA and represents the most frequent cause of premature death. Most individuals begin experiencing initial symptoms between 10 and 15 years of age, although there are atypical late-onset forms with initial symptoms presenting after age 25.

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 the FXN gene. 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 and 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. However, 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.

INTERPRETATION

Normal results (≥15 ng/mL for pediatric and ≥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.

REFERENCE VALUES

Pediatric (<18 years) normal frataxin: ≥15 ng/mL

Adults (≥18 years) normal frataxin: ≥21 ng/mL

ANALYTIC TIME

14 days

CONTENT AND VALUES SUBJECT TO CHANGE. SEE THE MAYO MEDICAL LABORATORIES TEST CATALOG FOR CURRENT INFORMATION.
CLINICAL REFERENCE

