Lab Dept: Anatomic Pathology

Test Name: FRIEDREICH ATAXIA (FRDA)

General Information

Lab Order Codes: FRDA

Synonyms: Gene X25 Mutation; FRDA

CPT Codes: 81401 – Molecular Pathology procedure, Level 2

Test Includes: Long PCR analysis across the GAA region of the FXN gene to determine allele sizes. All normal sized alleles and the vast majority of expanded alleles will be detected in this analysis. Southern analysis is used to identify extremely large GAA repeat expansions and to confirm the PCR results. Allele sizes will be reported as approximations and size ranges are subject to change as more information becomes available.

Logistics

Test Indications: Friedreich ataxia (FRDA) is an autosomal recessive neurodegenerative disorder with an incidence of approximately 1 in 50,000. FRDA is characterized by progressive gait and limb ataxia, lack of tendon reflexes in the legs, loss of position sense, dysarthria, and pyramidal weakness of the legs. Hypertrophic cardiomyopathy is found in almost all patients and diabetes mellitus is seen in 10% of the patients. The age of onset is usually around puberty, almost always before 25 years with a slow progression of the disease. The mutation responsible for FRDA resides in a novel gene (X25) which maps to 9q13 and is characterized by expansion of an unstable GAA trinucleotide repeat. In approximately 95% of cases normal individuals have 7-34 repeats, while FRDA patients have expansions off 66 repeats or greater.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Baylor College of Medicine, Medical Genetics Laboratory (Test#: 6031)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Sunday – Thursday, 24 hours

Turnaround Time: 3 weeks

Special Instructions:	Please fill out Baylor DNA (Non-sequencing order form) available online and submit to the laboratory with the specimen. It is also recommended that a consent form be submitted.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	3 - 5 mL blood (Adults/Children) 3 mL blood (Infants)
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture. Mix specimen thoroughly by gentle inversion.
Special Processing:	Lab Staff: **Do Not** centrifuge. Ship whole blood specimen in original collection container at room temperature via overnight courier. Do not heat or freeze. The sample must arrive at Baylor College on a weekday (Monday through Friday).
Patient Preparation:	None
Sample Rejection:	Specimens older than 48 hours, mislabeled or unlabeled specimens

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A
Limitations:	Due to limitations in methodology, alleles 35 to 60 repeats may not be distinguished from those under 35 repeats.
Methodology:	PCR, Southern Blot
References:	Baylor College of Medicine, Department of Molecular and Human Genetics March 2010
Updates:	2/5/2013: CPT update