

UCLA DIAGNOSTIC MOLECULAR PATHOLOGY LABORATORY
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Friedreich's Ataxia Mutation Analysis

↑CPT

83890; 84311; 83898; 83894; 83912

↑Synonyms

FRDA Gene; Trinucleotide Repeat Disorder

↑Test Includes

Sizing the GAA trinucleotide repeat expansion in the FRDA gene.

↑Laboratory

Molecular Pathology

↑Availability

Monday-Friday, 0700-1700

↑Turnaround Time

3-14 days

↑Specimen

Whole blood or amniotic fluid

↑Volume

4 mL

↑Container

Lavender top (EDTA) tube

↑ Storage Instructions

All specimens should be sent to the Laboratory immediately after collection, preferably by overnight delivery. Specimens should be kept at room temperature or refrigerated but not frozen.

↑ Causes for Rejection

Blood samples frozen and thawed will yield low quality DNA; specimens inadequately identified

↑ Reference Range

Normal GAA trinucleotide repeat lengths are between 5 and 33.

↑ Use

Friedreich ataxia, an autosomal recessive spinocerebellar degeneration which affects both the central and peripheral nervous systems, presents with progressive nerve conduction loss, hyporeflexia, ataxia, dysarthria, and movement disorder beginning before adolescence. An associated hypertrophic cardiomyopathy is the cause of death in many cases. The cloning of the FRDA gene demonstrates a clear correlation between the clinical diagnosis and a GAA triplet expansion in intron 1. Campuizano et al found homozygous triplet repeats in 81/84 patients. The three remaining patients were heterozygous for an expanded GAA repeat on one allele and a point mutation in the coding region of the gene on the other allele. Follow-up papers show a correlation between severity and the length of the GAA repeats.

↑ Limitations

Point mutations within the FRDA gene which are found in a small percentage of FA patients and carriers, will not be detectable by this test. It is also possible that an extremely large GAA expansion may not be detectable because of limitations in PCR amplification.

↑ Methodology

FRDA genotyping involves the isolation of DNA in peripheral blood and amplification of specific regions of the FRDA gene by Polymerase Chain Reaction (PCR). The PCR products for the FRDA gene create specific electrophoretic banding patterns allowing sizing of the GAA expansion.
