

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** Friedreich Ataxia Mutation Analysis
- **Test Code:** 2419

Related Diseases & Genes:

- Friedreich ataxia with retained reflexes (OMIM #: 229300) || Gene: *FXN* (OMIM #: 606829)
- Friedreich ataxia (OMIM #: 229300)

Synonyms:

- FRIEDREICH ATAXIA 1
- FRDA1
- FXN Gene Mutation Analysis
- Frataxin Mutation
- FA
- X25 Gene Mutation Analysis

Test Details:

- **Methodology:** PCR
- **Performed:** Everyday
- **Reported:** 2 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Neurogenetics

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