| Patient ID | Patient Name | | Birth Date | Gender | Age |
|---|---|---|---|---|---|
| SA00150321 | TESTINGRNV, REPORTVALIDATION | | 2021-12-12 | F | 6 W |
| **Order Number** | **Client Order Number** | **Ordering Physician** | **Report Notes** | | |
| SA00150321 | SA00150321 | CLIENT,CLIENT | | | |
| **Account Information** | | **Collected** | | | |
| C7028846 DLMP Rochester | | 24 Jan 2022 20:00 | | | |

# FXN, Repeat Expansion Analysis

## Result Summary

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NEGATIVE

## Result

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Number of GAA repeats: 5 (Normal) and 7 (Normal)

## Interpretation

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This result decreases the likelihood that this individual has Friedreich ataxia (FA). A diagnosis of FA cannot be completely excluded as other alterations within the FXN gene, such as single nucleotide variants or deletions, are not detected by this method.

If a diagnosis is still suspected, consider obtaining a frataxin level in blood (FFRWB / Frataxin, Quant, WB). Additionally, testing the FXN gene via alternative methodologies can be used to identify variants not detected by this assay. Please contact the Genomics Laboratory at 1–800–533–1710 with questions about these tests.

A genetic consultation may be of benefit.

## Reason for Referral

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Evaluation for a diagnosis of Friedreich ataxia. Test for the presence of a GAA repeat expansion in the FXN gene.

## Specimen

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WB Whole Blood

## Method

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PCR-based methods were utilized to detect GAA trinucleotide repeat expansions within the FXN gene.

**Normal:** <34
**Borderline:** 34–65
**Expanded:** >65

## Disclaimer

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Cautions
For familial testing, it is important to first document the molecular etiology of disease in an affected family member to confirm that a repeat expansion is the underlying mechanism of disease in the family. Specifically, this assay will not detect nonrepeat expansion variants (eg, sequence variants, deletions, and duplications).

It is strongly recommended that patients undergoing genetic testing receive genetic counseling.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data, such as frataxin concentrations (see FFRWB / Friedreich Ataxia, Frataxin, Quantitative, Blood and FFRBS / Friedreich Ataxia, Frataxin, Quantitative, Blood Spot). Errors in test interpretation may occur if the provided information is inaccurate or incomplete.

Rare variants (ie, polymorphisms) may exist, such as intron 1 deletions, which could lead to false-negative results. If GAA-repeat expansion results do not match clinical findings, additional testing should be considered.

Due to somatic mosaicism, GAA repeat-sizes in peripheral blood specimens may not reflect GAA repeat-sizes in other tissues (eg, central nervous system).

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories at 800–533–1710 for instructions for testing patients who have received a bone marrow transplant.

## Released By

MCR

Devin Oglesbee, Ph.D.

**Received:** 25 Jan 2022 11:22 **Reported:** 01 Feb 2022 17:11



**Performing Site Legend**

| Code | Laboratory | Address | Lab Director | CLIA Certificate |
|---|---|---|---|---|
| MCR | Mayo Clinic Laboratories - Rochester Main Campus | 200 First Street SW, Rochester, MN 55905 | William G. Morice M.D. Ph.D | 24D0404292 |