

Patient ID SA00150327	Patient Name TESTINGRNV, REPORTVALIDATION	Birth Date 2021-11-08	Gender M	Age 2 M
Order Number SA00150327	Client Order Number SA00150327	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Jan 2022 20:00		

FXN, Repeat Expansion Analysis

Result Summary

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POSITIVE

Result

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Number of GAA repeats: 600 (Full Penetrance) and 1000 (Full Penetrance)

Interpretation

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This result is consistent with a molecular diagnosis of Friedreich ataxia (FA). Of note, severity of symptoms and age of onset are correlated with the GAA repeat size of the smaller allele, with a larger number of GAA repeats corresponding to increased severity of symptoms and earlier age of onset of disease. Of note, this assay cannot rule out the possibility of larger repeat sizes in other tissues, such as the central nervous system, due to somatic mosaicism.

As full penetrance alleles were detected in this individual, genetic testing of at risk family members could be considered. If appropriate, genetic testing should be offered to this individual's reproductive partner to further clarify the risk of having a child affected with FA.

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.

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



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Released By

MCR

Devin Oglesbee, Ph.D.

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