

Patient ID SA00100692	Patient Name TESTINGRNV, REPORT	Birth Date 1989-01-17	Gender F	Age 28
Order Number SA00100692	Client Order Number SA00100692	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Nov 2017 14:00		

C9orf72, Molecular Analysis

Result Summary

POSITIVE

Result

GGGGCC repeat: >100 (Expanded) and 2

Interpretation

Given the family history reported and the expanded allele identified, this individual is at increased risk for developing a C9orf72-related disorder (c9FTD, c9ALS or c9FTD/ALS). Of note, an allele in this range may be associated with clinical heterogeneity (age of onset/ disease duration) and incomplete penetrance/age-dependent penetrance for C9orf72-related disorders. Therefore, the clinical significance of this result should be interpreted within the context of the family history and clinical presentation of this individual.

Identification of a mutation has important implications for family members. Testing of at risk individuals is possible, but it is recommended that predictive testing be performed in conjunction with appropriate pre- and post-test counseling.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or

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false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

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

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Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Reason for Referral

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Family history of frontotemporal dementia (FTD) and/or amyotrophic lateral sclerosis (ALS). Test for the presence of an expansion in the C9orf72 gene.

Specimen

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WB Whole Blood

Method

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A combined amplicon-length and repeat-primed PCR-based assay was used to size alleles up to approximately 145 repeats and detect expansions of GGGGCC hexanucleotide repeats in the 5' non-coding region of the C9orf72 gene (NM_001256054). The reported number of GGGGCC repeats for the PCR based assay is estimated to be correct within $\pm 5\%$.

Normal: <20

Indeterminate: 20–100

Positive: >100

Released By

MCR

Cristiane M. Ida, M.D.

Received: 13 Nov 2017 15:38

Reported: 14 Nov 2017 10:23

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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Test
Environment
ETBM Template

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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