

Patient ID SA00001251	Patient Name SAMPLEREPDMGLM, VLD20150727A0001	Birth Date 1981-01-01	Gender M	Age 34
Order Number SA00001251	Client Order Number SA00001251	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 26 Jul 2015 14:27		

DRPLA Gene Analysis

Result Summary

EXPANDED MUTATION IDENTIFIED

MCR

Result

CAG repeat: 60 (Expanded) and 13

MCR

Interpretation

1 MCR

This result is consistent with a diagnosis of or predisposition to dentatorubral-pallidoluysian atrophy (DRPLA).

Identification of an expansion in the ATN1 gene has important implications for family members. Predictive testing for at risk family members should be performed in conjunction with appropriate pre- and post-testing counseling.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

A PCR-based assay was utilized to detect CAG repeat expansions within the ATN1 gene. We estimate that the number of CAG repeats is correct to within $\pm 5\%$. Normal: 7–35; Expanded: 49–93 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 false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

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.

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.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

Received: 28 Jul 2015 08:22

Reported: 28 Jul 2015 11:54

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.

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