MAYO CLINIC LABORATORIES

1-800-533-1710

# DRPL

Dentatorubral-Pallidoluysian Atrophy (DRPLA) Gene Analysis, Varies

| Patient ID | Patient Name | | Birth Date | Gender | Age |
|---|---|---|---|---|---|
| SA00000939 | SAMPLEREPDMGLM, VLD20150713A0200 | | 1981-01-01 | M | 34 |
| Order Number | Client Order Number | Ordering Physician | Report Notes | | |
| SA00000939 | SA00000939 | CLIENT, CLIENT | | | |
| Account Information | | Collected | | | |
| C7028846 DLMP Rochester | | 12 Jul 2015 17:37 | | | |

## DRPLA Gene Analysis

### Result Summary

MCR

NEGATIVE

### Result

MCR

CAG repeat: 10 and 13 (Normal)

### Interpretation

[1] MCR

This result suggests that this individual is very unlikely to have dentatorubral-pallidoluysian atrophy (DRPLA). To date, alterations other than expansions within the ATN1 gene have not been associated with DRPLA. This result does not rule out the diagnosis of other inherited neurodegenerative disorders that have overlapping clinical features with DRPLA.

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 ±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:** 13 Jul 2015 20:26 **Reported:** 23 Jul 2015 14:59



### 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 |