**Test** | **Flag** | **Results** | **Unit** | **Reference Value** | **Perform Site**
---|---|---|---|---|---
DRPLA Gene Analysis
Result Summary | See Below | MCR
Result: EXPANDED MUTATION IDENTIFIED
Result | See Below | MCR
Result: CAG repeat: 60 (Expanded) and 13
Interpretation | 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 +/-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 Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.
Multiple in-silico evaluation tools may have been used to

***Performing Site Legend on Last Page of Report***

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**Patient Name** | **Collection Date and Time** | **Report Status**
---|---|---
SAMPLEREPDMGLM,VLD20150727A0001 | 07/26/2015 14:27 | Final

* Report times for Mayo performed tests are CST/CDT
Laboratory Service Report

Patient Name
SAMPLEREPMGLM,VLD20150727A0001

Patient ID
SA00001251

Age
34

Gender
M

Order #
SA00001251

DOB
01/01/1981

Ordering Phys
CLIENT,CLIENT

Client Order #
SA00001251

Collected
07/26/2015 14:27

Printed
07/30/2015 10:22

Test
Flag
Results
Unit
Reference
Value
Perform
Site*

assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Laboratory developed test.
PDF Report available at:
https://test.mmlaccess.com/Reports/C7028846-x0N2YnXTJx.ashx

Specimen
WB Whole Blood

Released By
EMILY LAUER

RECEIVED: 07/28/2015 08:22
REPORTED: 07/28/2015 11:54

* Performing Site:
MCR Mayo Clinic Laboratories - Rochester Main Campus
200 First St SW Rochester, MN 55905
Lab Director: William G. Morice, II, M.D., Ph.D.

Patient Name
SAMPLEREPMGLM,VLD20150727A0001

Collection Date and Time
07/26/2015 14:27

Report Status
Final

** End of Report **

* Report times for Mayo performed tests are CST/CDT
RESULT SUMMARY
EXPANDED MUTATION IDENTIFIED

RESULT
CAG repeat: 60 (Expanded) and 13

INTERPRETATION
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.

SPECIMEN
WB Whole Blood

METHOD
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

DISCLAIMER
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determination of pathogenicity is currently unvalidated.

Laboratory developed test.

REleased by
EMILY LAUER

<table>
<thead>
<tr>
<th>CODE</th>
<th>LABORATORY</th>
<th>ADDRESS</th>
<th>LAB DIRECTOR</th>
</tr>
</thead>
<tbody>
<tr>
<td>MCR</td>
<td>Mayo Clinic Laboratories - Rochester Main Campus</td>
<td>200 FIRST STREET SW ROCHESTER MN 55905-0001</td>
<td>WILLIAM G MORICE, II, MD, PhD</td>
</tr>
</tbody>
</table>

Report times for Laboratory Name performed tests are CST/CDT.
The collected, received, and reported dates and times on the report are in the time zone of the performing location.