MAYO CLINIC LABORATORIES

1-800-533-1710

HAD

Huntington Disease, Molecular Analysis, Varies

Patient ID	Patient Name		Birth Date	Gender	Age
SA00000808	SAMPLEREPDMGLM, VLD20150713A0068		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00000808	SA00000808	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 14:55			

Huntington Disease Analysis

Result Summary

MCR

FULL MUTATION IDENTIFIED

Result

MCR

CAG repeat: 45 (Full penetrance) and 13

Interpretation

[1] MCR

This result is consistent with a diagnosis of Huntington disease.

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

A PCR-based assay was utilized to detect CAG repeat expansions in exon 1 of the HTT gene. We estimate that the number of CAG repeats is correct within ±5%.

Normal: <27
Intermediate: 27–35
Reduced penetrance: 36–39
Full penetrance: >39
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.

Reason for Referral

MCR

Evaluation for a diagnosis of Huntington disease (HD). Test for the presence of an expansion in the HTT gene.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:20 **Reported:** 24 Jul 2015 12:47

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