

Patient ID <b>SA00000966</b>	Patient Name <b>SAMPLEREPDMGLM, VLD20150713A0227</b>	Birth Date <b>1981-01-01</b>	Gender <b>M</b>	Age <b>34</b>
Order Number <b>SA00000966</b>	Client Order Number <b>SA00000966</b>	Ordering Physician <b>CLIENT, CLIENT</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>12 Jul 2015 18:08</b>		

## Huntington Disease Analysis

### Result Summary

**NEGATIVE**

### Result

CAG repeat: 12 and 10 (Normal)

### Interpretation

This result suggests that this individual is very unlikely to have Huntington disease (HD). To date, alterations other than CAG expansions in the HTT gene have not been associated with HD. This result does not rule out the diagnosis of other inherited neurodegenerative disorders that have overlapping clinical features with HD.

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  $\pm 5\%$ .

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

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ClinGen. This may not be applicable for all tests.

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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.

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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

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

### Specimen

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WB Whole Blood

### Released By

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EMILY LAUER

**Received:** 13 Jul 2015 20:27

**Reported:** 24 Jul 2015 18:47

### 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