

Client: Example Client ABC123  
123 Test Drive  
Salt Lake City, UT 84108  
UNITED STATES

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 11/15/2000  
**Gender:** Unknown  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Huntington Disease (HD) CAG Repeat Expansion**

ARUP test code 3016908

Huntington Disease Specimen whole Blood

Huntington Disease Allele 1 41 CAG Repeats

Huntington Disease Allele 2 20 CAG Repeats

Huntington Disease Interpretation See Note

Indication for Testing: Diagnostic or predictive testing for Huntington disease (HD).

Interpretation: This individual has one allele in the normal range and one allele in the full penetrance affected range. He/she is predicted to develop Huntington disease sometime within a normal lifespan. This individual's offspring have a 50% risk for inheriting an expanded allele and being affected with HD.

Recommendations: Genetic consultation is recommended.

This result has been reviewed and approved by [REDACTED]

**H=High, L=Low, \*=Abnormal, C=Critical**

**BACKGROUND INFORMATION:** Huntington Disease (HD) CAG Repeat Expansion

**CHARACTERISTICS:** Neurodegenerative disorder causing progressive cognitive, motor, and psychiatric disturbances typically beginning at 35-44 years of age. An estimated 5 percent of individuals with HD are symptomatic as juveniles and 25 percent of individuals after age 50.

**INCIDENCE:** 1 in 15,000.

**INHERITANCE:** Autosomal dominant.

**CAUSE:** Expanded number of CAG repeats in the HTT gene. HD allele with reduced penetrance 36-39 CAG repeats; HD allele with full penetrance 40 or more CAG repeats.

**CLINICAL SENSITIVITY AND SPECIFICITY:** 99 percent.

**METHODOLOGY:** Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary electrophoresis. Repeat sizing precision is +/- 2 for alleles less than or equal to 50 repeats, +/- 3 for alleles with 51 to 75 repeats, and +/- 4 for alleles greater than 75 repeats.

**ANALYTIC SENSITIVITY AND SPECIFICITY:** 99 percent.

**LIMITATIONS:** Other neurodegenerative disorders will not be detected. Diagnostic errors can occur due to rare sequence variations. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation.

Phenotype	Number of CAG Repeats
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Normal allele	less than or equal to 26
Mutable normal (intermediate) allele	27-35
HD allele with reduced penetrance	36-39
HD allele with full penetrance	greater than or equal to 40

**COMPLIANCE STATEMENT:** Laboratory Developed Test (LDT)/Genetic

**VERIFIED/REPORTED DATES**

Procedure	Accession	Collected	Received	Verified/Reported
Huntington Disease Specimen	23-317-155680	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Allele 1	23-317-155680	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Allele 2	23-317-155680	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Interpretation	23-317-155680	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

**END OF CHART**

**H=High, L=Low, \*=Abnormal, C=Critical**

Unless otherwise indicated, testing performed at: