

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 8/10/1959  
**Gender:** Female  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Huntington Disease (HD) Mutation by PCR**

ARUP test code 0040018

Huntington Disease (HD) - Specimen whole Blood

Huntington Disease (HD) Allele 1 44 CAG Repeats

Huntington Disease (HD) Allele 2 17 CAG Repeats

Huntington Disease (HD) Interpretation See Note

This individual has one allele in the normal range and one allele in the affected range. He/she is predicted to develop Huntington Disease (HD) sometime within a normal lifespan. His/her offspring have a 50% risk for inheriting an expanded HD allele and being affected with HD. Genetic Consultation is recommended.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

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

Background Information for Huntington Disease (HD) Mutation by PCR:

Characteristics: Neurodegenerative disorder causing progressive cognitive, motor, and psychiatric disturbances typically beginning at 35-44 years of age (Although, 5 percent are affected as juveniles and 25 percent affected after age 50).  
 Incidence: 1 in 15,000.  
 Inheritance: Autosomal dominant.  
 Cause: Expanded number of CAG repeats in the HD gene (27-35 repeats-unaffected, intermediate; 36-39 repeats-reduced penetrance; 40+ repeats-affected, full penetrance).  
 Clinical Sensitivity and Specificity: 99 percent.  
 Methodology: Chimeric PCR followed by size analysis using capillary electrophoresis.  
 Analytical Sensitivity and Specificity: 99 percent.  
 Limitations: Other neurodegenerative disorders will not be detected. Rare, previously unreported variants may interfere with PCR amplification. Diagnostic errors can occur due to rare sequence variations.

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

Counseling and informed consent are required for Huntington Disease genetic testing. Consent forms are available online at www.aruplab.com.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Huntington Disease (HD) - Specimen	19-154-111802	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease (HD) Allele 1	19-154-111802	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease (HD) Allele 2	19-154-111802	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease (HD) Interpretation	19-154-111802	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