

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 6/9/1948
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 17 CAG Repeats

Huntington Disease (HD) Allele 2 16 CAG Repeats

Huntington Disease (HD) Interpretation See Note

This individual has two normal alleles, therefore, will neither be affected with, nor transmit, Huntington disease.
This result has been reviewed and approved by Hunter Best, 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	19-149-132036	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease (HD) Allele 1	19-149-132036	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease (HD) Allele 2	19-149-132036	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease (HD) Interpretation	19-149-132036	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