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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/1752
Sex:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Huntington Disease (HD) Mutation by PCR (Extended TAT as of 11/20/20-no referral available)

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

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]

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. 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.
Analytical 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.

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

Phenotype	Number of CAG Repeats
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

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Huntington Disease (HD) - Specimen	22-137-109352	5/17/2022 12:38:00 PM	5/17/2022 12:38:13 PM	5/17/2022 12:47:00 PM
Huntington Disease (HD) Allele 1	22-137-109352	5/17/2022 12:38:00 PM	5/17/2022 12:38:13 PM	5/17/2022 12:47:00 PM
Huntington Disease (HD) Allele 2	22-137-109352	5/17/2022 12:38:00 PM	5/17/2022 12:38:13 PM	5/17/2022 12:47:00 PM
Huntington Disease (HD) Interpretation	22-137-109352	5/17/2022 12:38:00 PM	5/17/2022 12:38:13 PM	5/17/2022 12:47:00 PM

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