ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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	28 CAG Repeats
Huntington Disease Allele 2	16 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 mutable normal (intermediate) range. Thus, this individual is not at risk for developing Huntington disease (HD) but may transmit an expanded allele to offspring. Expansion risk is influenced by the transmitting individual's sex and CAG repeat size. Males with an intermediate allele of 27-33 CAG repeats have a less than 1 percent risk of transmitting a potentially disease-causing expanded allele to their offspring (see Semaka 2013; Semaka and Hayden 2014). Females with an intermediate range allele have not been observed to have offspring with HD.

Recommendations: Genetic consultation is recommended.

References:
Semaka, A. et al. CAG size-specific risk estimates for intermediate allele repeat instability in Huntington disease. J Med Genet. 2013; 50:696-703.

Semaka, A. and Hayden, M.R. Evidence-based genetic counselling implications for Huntington disease intermediate allele predictive test results. Clin Genet. 2014; 85:303-11.

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 SENSITIVITYE 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
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	23-319-149125	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Allele 1	23-319-149125	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Allele 2	23-319-149125	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Interpretation	23-319-149125	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