



LABORATORY REPORT

QC ACCOUNT (WARDE)
300 W. TEXTILE
ANN ARBOR MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 46 Y

Referral Testing

Collected: 08/13/2024 15:12 Received: 08/13/2024 15:12

Table with 6 columns: Test Name, Result, Flag, Ref-Ranges, Units, Site. Row 1: Huntington Disease (HD) CAG Repeat Expansion. Row 2: Huntington Disease Specimen, Whole Blood, ARRL. Row 3: Huntington Disease Allele 1, 15, CAG Repeats, ARRL. Row 4: Huntington Disease Allele 2, 16, CAG Repeats, ARRL. Row 5: Huntington Disease Interpretation, See Note, ARRL.

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.
METHODODOLOGY: 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.

Table with 2 columns: Phenotype, Number of CAG Repeats. Row 1: Normal allele, less than or equal to 26. Row 2: Mutable normal (intermediate) allele, 27-35. Row 3: HD allele with reduced penetrance, 36-39. Row 4: HD allele with full penetrance, greater than or equal to 40.

COMPLIANCE STATEMENT: Laboratory Developed Test (LDT)/Genetic
Performed By: ARUP Laboratories
500 Chipeta Way
Salt Lake City, UT 84108
Laboratory Director: Jonathan R. Genzen, MD, PhD

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED



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CLIA Number: 46D0523979

Reported Date: 08/13/2024 15:12 HDCAG

Performing Site:

ARRL: ARUP REFERENCE LAB 500 Chipeta Way Salt Lake City UT 841081221

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

G413000043
WX0000003827

Ordered By: KAJAL SITWALA, MD, PHD
WX00000000002516

Printed D&T: 08/13/24 15:13

Kajal V. Sitwala, MD, PhD - Medical Director

Form: MM RL1

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