Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 10/20/2023 11:53 Received: 10/20/2023 11:53

Test Name	Result	Flag	Ref-Ranges	Units	Site
Huntington Disease (HD) CAG Repeat Expansion					
Huntington Disease Specimen	Whole Blood				ARRL
		Reported Date:	2023.10.20	-:1	
Huntington Disease Allele 1	15			CAG Repeats	ARRL
		Reported Date:	2023.10.20	-:1	
Huntington Disease Allele 2	16			CAG Repeats	ARRL
		Reported Date:	2023.10.20	-:1	
Huntington Disease Interpretation	See Note				ARRL

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

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

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Test Name	Result	Flag	Ref-Ranges	Units	Site
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
CLIA Number: 46D0523979

Reported Date: 2023.10.20 11:53 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

F420000010
WX0000003827
Printed D&T: 10/20/23 11:54

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002365

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1