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Patient Report

Patient Age/Sex: 1 day Male

Specimen Collected: 21-Mar-22 15:03

X-Huntington Disease Mutation by PCR | **Received: 21-Mar-22 15:03** | **Report/Verified: 21-Mar-22 15:27**

Procedure	Result	Units	Reference Interval
Huntington Disease (HD) -Specimen	Whole Blood		
Huntington Disease (HD) Allele 1	27	CAG Repeats	
Huntington Disease (HD) Allele 2	20	CAG Repeats	
Huntington Disease (HD) Interpretation	See Note [f1 i1]		

Result Footnote

f1: Huntington Disease (HD) Interpretation

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

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 Rong Mao, M.D.

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Test Information

i1: Huntington Disease (HD) Interpretation

Background Information for Huntington Disease (HD) Mutation by PCR:

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Test Information

i1: Huntington Disease (HD) Interpretation

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.

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.