HOTLINE: Effective May 16, 2022

0040018 **Huntington Disease (*HD*) Mutation by PCR (Extended TAT as of 11/20/20-no referral available)** **HD**

Specimen Required: Collect: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Preferred transport temp: Refrigerated. Also acceptable: Room temperature.
Remarks: A completed HD specific consent form, signed by the patient (or legal guardian) and physician, is required for all specimens. Testing for patients under the age of 18 years or fetal specimens is not offered. Presymptomatic patients are strongly encouraged to be tested through a counseling program approved by the Huntington Disease Society of America at (800) 345-4372. Call Genetics Processing with additional questions at 800-242-2787 ext 3301.
Stability (collection to initiation of testing): Room temperature: 1 week; Refrigerated: 1 month; Frozen: unacceptable

Interpretive Data:

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 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 ≤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.

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.

Allele type	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

HOTLINE NOTE: Remove information found in the Reference Interval field.