SCHOOL OF MEDICINE
INDIANA UNIVERSITY

Department of Medical and Molecular Genetics
Division of Diagnostic Genomics

Laboratory Test Directory

Huntington Disease Mutation by PCR

CPT Code(s): 81401

Service Code (IU Health): 53052494

Ordering Recommendation: Diagnostic confirmation in individuals with clinical symptoms of Huntington disease; presymptomatic testing for adults with a positive family history.

Synonyms: Huntington disease, Huntington chorea

Methodology: Triplet repeat primed PCR and capillary electrophoresis. Reflex Southern analysis is performed as a send-out test as needed to further characterize expanded/abnormal alleles.

Performed: Mon-Fri

Reported: 6-9 days

Specimen Requirements

Patient Preparation: None required for whole blood

Collect: Lavender (EDTA) tubes

Specimen Volume: Blood: 2-6 mL whole blood

Storage/Transport: Refrigerated/Room temperature

Unacceptable Conditions: Grossly hemolyzed or clotted

Remarks:

Stability: 2 weeks refrigerated; 1 month frozen

Reference Interval: by report

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Interpretive Data

Characteristics: Huntington disease (HD) is inherited as an autosomal dominant condition which affects the central nervous system. Neuron degeneration can cause uncontrolled movements, loss of intellectual faculties, and emotional disturbance in patients with HD. Some early clinical symptoms include dementia, mood swings, depression, lack of coordination, irritability or trouble driving and learning new things, remembering a fact, or making a decision. As the disease progresses, concentrating on intellectual tasks becomes increasingly more difficult. The symptoms and severity of the disease vary between individuals as well as the age of onset.

Inheritance: Autosomal dominant

Cause: CAG repeat expansion in the coding region of the *Huntingtin (HTT)* gene located at 4p16.3. The CAG repeat size varies from: <27 repeats (normal); 27-35 CAG repeats (mutable normal); 36-39 repeats (abnormal with reduced penetrance); ≥40 repeats (abnormal with full penetrance).

Incidence: 1:15000

Penetrance: Variable

Analytical sensitivity and specificity: 99%

Limitations: Only the CAG expansion will be detected. Mutations or variants will not be detected. Although rare, false positive or false negative results may occur. All results should be interpreted in context of clinical findings, relevant history, and other laboratory data.

References: Losekoot M et al. EMQN/CMGS best practice guidelines for the molecular genetic testing of Huntington disease. European Journal of Human Genetics (2012), 1–7.