

Huntington Disease: CAG Repeat Analysis

Test Code: HT

Turnaround time: 2 weeks

CPT Codes: 81401 x1

Condition Description

Huntington disease is a progressive neurodegenerative disease characterized by chorea and dementia. This disease is inherited in an autosomal dominant fashion and affects approximately 1 per 10,000 individuals. Although patients may develop symptoms at any time from childhood to later in life, the average age of onset is between 35 and 45. With paternal transmission of the gene, there is a tendency for earlier onset to occur. Over 80% of the patients with an onset before the age of 20 inherited the abnormal gene from their father. Mutations may occur spontaneously as well, but this is uncommon.

Huntington disease is caused by an abnormal expansion of a CAG repeat within the *HTT/IT15* on chromosome 4. Although the number of CAG repeats correlates with the age of onset, the correlation is not precise enough to use in determining disease prognosis for an individual patient.

Due to the complexities and ethical issues surrounding genetic testing for Huntington, **a signed consent form for Huntington disease testing is mandatory**. Please call our laboratory to review your patient's case in advance and obtain the necessary forms.

[Click here](#) for the GeneReviews summary on this condition.

Genes

HD, [HTT](#), IT15

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Huntington disease
- Presymptomatic testing in adults with a family history of Huntington disease

Methodology

CAG repeat number is determined by PCR amplification and fragment size analysis.

Detection

Nearly 100% of CAG expansions in the *HTT/IT15* gene will be detected by this assay.

Reference Range

Normal individuals have 6-35 repeats while individuals with greater than 39 CAG repeats have a high probability of developing the disease. When the repeat number is between 36 and 39, Huntington disease cannot be reliably diagnosed due to incomplete penetrance.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (EDTA)

Specimen Requirements:

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

Type: DNA, Isolated

Specimen Requirements:

Microtainer

20µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Special Instructions

A signed [consent form](#) for Huntington disease testing is mandatory.