Overview

Useful For
Molecular confirmation of clinically suspected cases of Huntington disease (HD)
Presymptomatic testing for individuals with a family history of HD and a documented expansion in the HTT gene

Special Instructions
- Informed Consent for Genetic Testing
- Molecular Genetics: Neurology Patient Information
- Informed Consent for Genetic Testing (Spanish)

Method Name
Polymerase Chain Reaction (PCR)

NY State Available
Yes

Specimen

Specimen Type
Varies

Shipping Instructions
Specimen preferred to arrive within 96 hours of draw.

Necessary Information
Molecular Genetics: Neurology Patient Information or a recent clinical note is required. Testing cannot proceed without this information. See Special Instructions.

Specimen Required
Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Specimen Type: Whole blood

Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.
**Forms**

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

   - [Informed Consent for Genetic Testing](T576)
   - [Informed Consent for Genetic Testing-Spanish](T826)

2. If not ordering electronically, complete, print, and send a [Neurology Specialty Testing Client Test Request](T732) with the specimen.

**Specimen Minimum Volume**

1 mL

**Reject Due To**

All specimens will be evaluated by Mayo Clinic Laboratories for test suitability.

**Specimen Stability Information**

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<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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**Clinical and Interpretive**

**Clinical Information**

Huntington disease (HD) is an autosomal dominant progressive neurodegenerative disorder caused by a CAG repeat expansion in the *HTT* gene. HD is associated with cognitive impairment leading to dementia and a wide range of neuropsychiatric problems including apathy, depression, anxiety, and other behavioral disturbances. Additionally, affected individuals typically develop extrapyramidal symptoms (eg, dystonia, dysarthria, chorea, gait disturbance, postural instability, oculomotor dysfunction).

**Reference Values**

Normal alleles: <27 CAG repeats

Intermediate alleles: 27-35 CAG repeats

Reduced penetrance: 36-39 CAG repeats

Full penetrance: >39 CAG repeats

An interpretive report will be provided.

**Interpretation**

An interpretive report will be provided.
Cautions

For predictive testing, it is important to first document the presence of a CAG-repeat amplification in the HTT gene in an affected family member to confirm that molecular expansion is the underlying mechanism of disease in the family.

We strongly recommend that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.

Predictive testing of an asymptomatic child is not recommended.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Clinical Reference


Performance

Method Description

A PCR-based assay is utilized to detect expansions of a CAG trinucleotide tract in exon 1 of the HTT gene. (Unpublished Mayo method)

PDF Report

No

Day(s) and Time(s) Test Performed

Monday, Thursday; Varies

Analytic Time

4 days upon receipt of sufficient clinical information for testing.

Maximum Laboratory Time

11 days upon receipt of sufficient clinical information for testing.

Specimen Retention Time

Whole Blood: 2 weeks (if available); Extracted DNA: 3 months

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.
**Test Classification**

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

**CPT Code Information**

81271-HTT (huntingtin) (eg, Huntington disease) gene analysis; evaluation to detect abnormal (eg, expanded) alleles

**LOINC® Information**

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