MAYO CLINIC LABORATORIES

Test Definition: HAD

Huntington Disease, Molecular Analysis, Varies

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 collection.

Necessary Information

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

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 whole blood specimen in original tube. **Do not aliquot**.

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:
-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

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Clinical & 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.

It is strongly recommended 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 the interpretation of results may occur if information given is inaccurate or incomplete.

Clinical Reference

1. ACMG/ASHG statement. Laboratory guidelines for Huntington disease genetic testing. The American College of Medical Genetics/American Society of Human Genetics Huntington Disease Genetic Testing Working Group. Am J Hum Genet. 1998 May;62(5):1243-1247
2. Potter NT, Spector EB, Prior TW: Technical standards and guidelines for Huntington disease testing. Genet Med. 2004;6(1):61-65. doi: 10.1097/01.gim.0000106165.74751.15

Performance

Method Description

A polymerase chain reaction-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) Performed

Wednesday

Report Available

7 to 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 & 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 account representative. For assistance, contact Customer Service.

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Test Classification

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

CPT Code Information

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

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
HAD	Huntington Disease Analysis	21763-8

Result ID	Test Result Name	Result LOINC® Value
52885	Result Summary	50397-9
52886	Result	53782-9
52887	Interpretation	69047-9
52888	Reason for Referral	42349-1
52889	Specimen	31208-2
52890	Source	31208-2
52891	Released By	18771-6