MAYO CLINIC
Mayo Medical Laboratories

Test Definition: HD

Huntington Disease, Molecular Analysis

Reporting Title: Huntington Disease Analysis
Performing Location: Rochester

Specimen Requirements:

Specimen must arrive within 96 hours of draw.

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. Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) in Special Instructions
2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
51201	Specimen	Alphanumeric		N/A
51202	Specimen ID	Alphanumeric		N/A
51203	Source	Alphanumeric		N/A
51204	Order Date	Alphanumeric		N/A
51205	Reason for Referral	Alphanumeric		N/A
51206	Method	Alphanumeric		N/A
51207	Result	Alphanumeric		N/A
51208	Interpretation	Alphanumeric		N/A
51209	Amendment	Alphanumeric		N/A

Result ID	Reporting Name	Type	Unit	LOINC®
51210	Review By:	Alphanumeric		N/A
51211	Release Date	Alphanumeric		N/A

CPT Code Information:

81401- HTT (huntingtin) (eg, Huntington disease), evaluation to detect abnormal (eg, expanded) alleles

For nonparticipating payers:
83891-Isolation or extraction of highly purified nucleic acid
83898 x 2-Amplification, target, each nucleic acid sequence
83909-Separation and identification by high-resolution technique
83912-Interpretation and report

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.