MAYO CLINIC LABORATORIES

# Test Definition: HAD

Huntington Disease, Molecular Analysis, Varies

**Reporting Title:** Huntington Disease Analysis
**Performing Location:** Rochester

## 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 Requirements:

**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 Type | Temperature | Time | Special Container |
|---|---|---|---|
| Varies | Ambient (preferred) | | |
| | Frozen | | |
| | Refrigerated | | |

## Result Codes:

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

**LOINC® and CPT codes are provided by the performing laboratory.**

### Supplemental Report:

No

### CPT Code Information:

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

### 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.