

Varies

Reporting Title: DRPLA Gene Analysis **Performing Location:** Rochester

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Specimen Type: Whole blood

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.

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. Molecular Genetics: Neurology Patient Information

3. 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	Туре	Unit	LOINC®
53234	Result Summary	Alphanumeric		50397-9
53235	Result	Alphanumeric		49631-5



Test Definition: DRPL

Dentatorubral-Pallidoluysian Atrophy (DRPLA) Gene Analysis, Varies

Result ID	Reporting Name	Туре	Unit	LOINC®
53236	Interpretation	Alphanumeric		69047-9
53237	Specimen	Alphanumeric		31208-2
53238	Source	Alphanumeric		31208-2
53239	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81177-ATN1 (ataxin 2) (eg, denatatorubral-pallidolyuysian atrophy) gene analysis, evaluation to detect abnormal (eg, expanded) alleles

Reference Values:

Normal alleles: 7-35 CAG repeats Abnormal alleles: 49-93 CAG repeats

An interpretive report will be provided.