Test Definition: DRPLA

Dentatorubral-Pallidoluysian Atrophy (DRPLA) Gene Analysis

Reporting Title: DRPLA Gene Analysis
Performing Location: Rochester

Specimen Requirements:

Specimen must arrive within 96 hours of draw.

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.
3. If not ordering electronically, submit a Molecular Genetics Request Form (Supply T245) with the specimen.

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
16889	Specimen	Alphanumeric		31208-2
16890	Specimen ID	Alphanumeric		In Process
16891	Source	Alphanumeric		N/A
16892	Order Date	Alphanumeric		N/A
16893	Reason For Referral	Alphanumeric		42349-1
16894	Method	Alphanumeric		49549-9
16895	Result	Alphanumeric		21756-2
16896	Interpretation	Alphanumeric		69047-9
16897	Amendment	Alphanumeric		In Process

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Result ID	Reporting Name	Type	Unit	LOINC®
16898	Reviewed By:	Alphanumeric		In Process
16899	Release Date	Alphanumeric		N/A

CPT Code: 1 × 81401

Reference Values:

Normal alleles: 7-35 CAG repeats
Abnormal alleles: 49-93 CAG repeats
An interpretive report will be provided.