Test Definition: C9ORF

C9orf72 Hexanucleotide Repeat, Molecular Analysis, Varies

Reporting Title: C9orf72, Molecular Analysis
Performing Location: Rochester

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

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 specimen in original tube.

Specimen Minimum Volume:

1 mL

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		

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Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
52852	Result Summary	Alphanumeric		50397-9
52853	Result	Alphanumeric		77635-1
52854	Interpretation	Alphanumeric		69047-9
52855	Reason for Referral	Alphanumeric		42349-1
52856	Specimen	Alphanumeric		31208-2
55158	Method	Alphanumeric		85069-3
52857	Source	Alphanumeric		31208-2
52858	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81479

Reference Values:

Normal alleles (reference):<20 GGGGCC repeats
Indeterminate alleles: 20-100 GGGGCC repeats
Pathogenic alleles: >100* GGGGCC repeats

*The exact cutoff for pathogenicity is currently undefined. Although additional studies are needed to confirm if 100 repeats is the cutoff for pathogenicity, most individuals affected with a C9orf72-related disorder have C9orf72 hexanucleotide repeat expansions with hundreds to thousands of repeats.

An interpretive report will be provided.