Test Definition: C9OR

C9orf72 Hexanucleotide Repeat, Molecular Analysis

Reporting Title: C9orf72, Molecular 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.

Specimen Type	Temperature	Time
Varies	Varies	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
51775	Reason for Referral	Alphanumeric		In Process
51776	Result	Alphanumeric		In Process
51777	Interpretation	Alphanumeric		In Process
51778	Method	Alphanumeric		In Process
51779	Amendment	Alphanumeric		In Process
51780	Specimen	Alphanumeric		In Process
51781	Source	Alphanumeric		In Process
51782	Reviewed By	Alphanumeric		In Process
51783	Release Date	Alphanumeric		In Process

CPT Code Information:

81479-Unlisted Molecular Pathology procedure
C9orf72, Follow Up Analysis
81479-Unlisted Molecular Pathology procedure

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
C9OFU	C9orf72, Follow Up Analysis			No	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
C9OFU	51767	Result	Alphanumeric		In Process
C9OFU	51768	Amendment	Alphanumeric		In Process
C9OFU	51769	Specimen	Alphanumeric		In Process
C9OFU	51770	Source	Alphanumeric		In Process
C9OFU	51771	Reviewed By	Alphanumeric		In Process
C9OFU	51772	Release Date	Alphanumeric		In Process

Reference Values:

Normal alleles: <20 GGGGCC repeats
Intermediate alleles: 20-29 GGGGCC repeats
Penetrance: >29* GGGGCC repeats

*Alleles greater than 30 repeats are outside the reportable range for this assay and are detected using the companion Southern blot assay. There is not enough information at this time to determine if 30 repeats is the cutoff for pathogenicity.

An interpretive report will be provided.