



C9orf72 Hexanucleotide Repeat, Molecular Analysis

Test ID: C9OR

USEFUL FOR:

- Molecular confirmation of clinically suspected cases of c9frontotemporal dementia (FTD)/amyotrophic lateral sclerosis (ALS)
- Presymptomatic testing for individuals with a family history of c9FTD/ALS and a documented expansion in the C9orf72 gene

METHODOLOGY: Polymerase Chain Reaction (PCR)

Penetrant, intermediate and homozygous alleles are confirmed by Southern blot.

REFERENCE VALUES: An interpretive report will be provided.

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.

NOTE:

- Molecular Genetics–Congenital Inherited Diseases Patient Information Sheet (Supply T521) in Special Instructions.
- 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 STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Varies	

CAUTIONS:

- For predictive testing, it is important to first document the presence of the hexanucleotide repeat amplification in the C9orf72 gene in an affected family member to confirm that molecular expansion is the underlying mechanism of disease in the family.
- We strongly recommend that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.
- Predictive testing of an asymptomatic child is not recommended.

- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

CPT CODE:

81479-Unlisted Molecular Pathology procedure

C9orf72, Follow Up Analysis

81479-Unlisted Molecular Pathology procedure

DAY(S) SET UP: Tuesday; 10 am

ANALYTIC TIME: 5 days

NOTE: The following referral test code(s) will become obsolete effective April 25, 2014.

Test Name	Test ID	Referral Lab Code	Referral Lab
C9ORF72 (FTD) DNA Test	ZW127	209	Athena Diagnostics
C9ORF72 DNA Test	ZW127	670	Athena Diagnostics

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710