Test Definition: AFTDP

Inherited Frontotemporal Dementia and Amyotrophic Lateral Sclerosis Gene Panel, Varies

Reporting Title: FTD and ALS Gene Panel
Performing Location: Rochester

Ordering Guidance:

First tier testing for a diagnosis of dementia or amyotrophic lateral sclerosis is C9ORF / C9orf72, Hexanucleotide Repeat, Molecular Analysis, Varies, which is included with this test but is also available separately.

Targeted testing for familial variants (also called site-specific or known mutations testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Customization of this panel and single gene analysis for any gene present on this panel are available. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

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: 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. Do not aliquot.
Specimen Stability Information: Ambient (preferred)/Refrigerated

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	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
617494	Test Description	Alphanumeric		62364-5
617495	Specimen	Alphanumeric		31208-2
617496	Source	Alphanumeric		31208-2
617497	Result Summary	Alphanumeric		50397-9
617498	Result	Alphanumeric		82939-0
617499	Interpretation	Alphanumeric		69047-9
618174	Additional Results	Alphanumeric		82939-0
617500	Resources	Alphanumeric		99622-3
617501	Additional Information	Alphanumeric		48767-8
617502	Method	Alphanumeric		85069-3
617503	Genes Analyzed	Alphanumeric		48018-6
617504	Disclaimer	Alphanumeric		62364-5
617505	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81403
81406 x 10
81404 x 3
81405 x 2
81407
81479
81479 (if appropriate for government payers)

Reference Values:

An interpretive report will be provided.

C9orf72 Repeats:
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 the cutoff for pathogenicity is 100 repeats, most individuals affected with a C9orf72-related disorder have C9orf72 hexanucleotide repeat expansions with hundreds to thousands of repeats.