Test Definition: MNDP

Inherited Motor Neuron Disease Gene Panel, Varies

Reporting Title: Motor Neuron Disease 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.

For individuals with both ALS and evidence of dementia, consider AFTDP / Inherited Frontotemporal Dementia and Amyotrophic Lateral Sclerosis Gene Panel, Varies,

Targeted testing for familial variants (also called site-specific or known variants 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 whole blood 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®
617650	Test Description	Alphanumeric		62364-5
617651	Specimen	Alphanumeric		31208-2
617652	Source	Alphanumeric		31208-2
617653	Result Summary	Alphanumeric		50397-9
617654	Result	Alphanumeric		82939-0
617655	Interpretation	Alphanumeric		69047-9
618186	Additional Results	Alphanumeric		82939-0
617656	Resources	Alphanumeric		99622-3
617657	Additional Information	Alphanumeric		48767-8
617658	Method	Alphanumeric		85069-3
617659	Genes Analyzed	Alphanumeric		48018-6
617660	Disclaimer	Alphanumeric		62364-5
617661	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Supplemental

CPT Code Information:

81443

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.