

Reporting Title: Congenital Myasthenia Gene Panel **Performing Location:** Rochester

Ordering Guidance:

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 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.



Inherited Congenital Myasthenic Syndrome Gene Panel, Varies

Specimen Type	Temperature	Time	Special Container	
Varies	Varies			

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
617520	Test Description	Alphanumeric		62364-5
617521	Specimen	Alphanumeric		31208-2
617522	Source	Alphanumeric		31208-2
617523	Result Summary	Alphanumeric		50397-9
617524	Result	Alphanumeric		82939-0
617525	Interpretation	Alphanumeric		69047-9
618176	Additional Results	Alphanumeric		82939-0
617526	Resources	Alphanumeric		99622-3
617527	Additional Information	Alphanumeric		48767-8
617528	Method	Alphanumeric		85069-3
617529	Genes Analyzed	Alphanumeric		48018-6
617530	Disclaimer	Alphanumeric		62364-5
617531	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.