

## Inherited Congenital Myasthenic Syndrome Gene Panel, Varies

**Test ID:** CMSP

**Explanation:** On the effective date, reflex test MATCC will be added. Formatting of the Specimen Required field will be standardized.

### Current Algorithm

For information see [Neuromuscular Myopathy Testing Algorithm](#)

### New Algorithm

For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge.

For information see [Neuromuscular Myopathy Testing Algorithm](#)

### Current Reflex Tests

None

### New Reflex Tests

Test ID	Reporting Name	Available Separately	Always Performed
MATCC	Maternal Cell Contamination, B	Yes	No

### Current 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.

### New 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. To modify this panel via CGPH, use the Neurologic Disorders disease state for step 1 on the [Custom Gene Ordering Tool](#).

Current Specimen Required
<p><b>Patient Preparation:</b> A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.</p> <p><b>Submit only 1 of the following specimens:</b></p> <p><b>Specimen Type:</b> Whole blood</p> <p><b>Container/Tube:</b></p> <p><b>Preferred:</b> Lavender top (EDTA) or yellow top (ACD)</p> <p><b>Acceptable:</b> None</p> <p><b>Specimen Volume:</b> 3 mL</p> <p><b>Collection Instructions:</b></p> <ol style="list-style-type: none"> <li>1. Invert several times to mix blood.</li> <li>2. Send whole blood specimen in original tube. <b>Do not aliquot.</b></li> </ol> <p><b>Specimen Stability Information:</b> Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days</p> <p><b>Additional Information:</b></p> <ol style="list-style-type: none"> <li>1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.</li> <li>2. To ensure minimum volume and concentration of DNA are met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.</li> </ol> <p><b>Specimen Type:</b> Saliva</p> <p><b>Patient Preparation:</b> Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.</p> <p><b>Supplies:</b></p> <p>DNA Saliva Kit High Yield (T1007)</p> <p>Saliva Swab Collection Kit (T786)</p> <p><b>Container/Tube:</b></p> <p><b>Preferred:</b> High-yield DNA saliva kit</p> <p><b>Acceptable:</b> Saliva swab</p> <p><b>Specimen Volume:</b> 1 Tube if using T1007 or 2 swabs if using T786</p> <p><b>Collection Instructions:</b> Collect and send specimen per kit instructions.</p> <p><b>Specimen Stability Information:</b> Ambient (preferred) 30 days/Refrigerated 30 days</p> <p><b>Additional Information:</b> Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When</p>

New Specimen Required
<p><b>Patient Preparation:</b> A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.</p> <p><b>Submit only 1 of the following specimens:</b></p> <p><b>Specimen Type:</b> Whole blood</p> <p><b>Container/Tube:</b> Lavender top (EDTA) or yellow top (ACD)</p> <p><b>Specimen Volume:</b> 3 mL</p> <p><b>Collection Instructions:</b></p> <ol style="list-style-type: none"> <li>1. Invert several times to mix blood.</li> <li>2. Send whole blood specimen in original tube. <b>Do not aliquot.</b></li> <li>3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information</li> </ol> <p><b>Specimen Stability Information:</b> Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days</p> <p><b>Additional Information:</b></p> <ol style="list-style-type: none"> <li>1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.</li> <li>2. To ensure minimum volume and concentration of DNA are met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.</li> <li>3. For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.</li> </ol> <p><b>Specimen Type:</b> Saliva</p> <p><b>Patient Preparation:</b> Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.</p> <p><b>Supplies:</b></p> <p>DNA Saliva Kit High Yield (T1007)</p> <p>Saliva Swab Collection Kit (T786)</p> <p><b>Container/Tube:</b></p> <p><b>Preferred:</b> High-yield DNA saliva kit</p> <p><b>Acceptable:</b> Saliva swab</p> <p><b>Specimen Volume:</b> 1 Tube if using T1007 or 2 swabs if using T786</p>

applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

**Specimen Type:** Extracted DNA

**Container/Tube:**

**Preferred:** Screw Cap Micro Tube, 2mL with skirted conical base

**Acceptable:** Matrix tube, 1mL

**Collection Instructions:**

1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
2. Include concentration and volume on tube.

**Specimen Stability Information:** Frozen (preferred) 1 year/Ambient/Refrigerated

**Additional Information:** DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants).

Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

**Collection Instructions:** Collect and send specimen per kit instructions.

**Specimen Stability Information:** Ambient (preferred) 30 days/Refrigerated 30 days

**Additional Information:** Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

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#### Current Retention Time

Whole blood: 30 days (if available); Extracted DNA: 3 months

#### New Retention Time

Whole blood: 28 days (if available); Saliva: 30 days (if available); Extracted DNA: 3 months

## Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.