

Test Definition: NMEM

Red Blood Cell Membrane Disorders Gene Panel, Next-Generation Sequencing, Varies

Reporting Title: RBC Membrane Sequencing, NGS

Performing Location: Rochester

Ordering Guidance:

Multiple hematology gene panels are available. For more information see NHHA and Subpanel Comparison Gene List.

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

Additional Testing Requirements:

This test is best interpreted in the context of protein studies and peripheral blood findings. Prior to sending this test, Coombs testing should be negative and consider evaluating a peripheral blood smear. This can be provided by ordering RBCME / Red Blood Cell Membrane Evaluation, Blood. Fill out the information sheet and indicate that a next-generation sequencing test was also ordered. Additionally, providing complete blood cell count data and clinical notes will allow more precise interpretation of results.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Necessary Information:

- 1. <u>Metabolic Hematology Next-Generation Sequencing (NGS) Patient Information</u> is required. Testing may proceed without the patient information; however, the information aids in providing a more thorough interpretation. Ordering providers are strongly encouraged to fill out the form and send with the specimen.
- 2. If form not provided, include the following information with the test request: clinical diagnosis, pertinent clinical history (ie, complete blood cell count results and relevant clinical notes), and differentials based on clinical or morphologic presentation.

Specimen Requirements:

Specimen Type: Whole blood

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.

Container/Tube:

Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD)
Specimen Volume: 3 mL
Collection Instructions:

- Invert several times to mix blood.
- 2. Send whole blood specimen in original tube. Do not aliquot.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated

Forms:



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- 1. Metabolic Hematology Next-Generation Sequencing (NGS) Patient Information (T816) is required.
- 2. **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)
- 3. If not ordering electronically, complete, print, and send a Benign Hematology Test Request (T755) with the specimen.

Specimen Type	Temperature	Time	Special Container	
Varies	Varies			

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
619062	Test Description	Alphanumeric		62364-5
619063	Specimen	Alphanumeric		31208-2
619064	Source	Alphanumeric		31208-2
619065	Result Summary	Alphanumeric		50397-9
619066	Result	Alphanumeric		82939-0
619067	Interpretation	Alphanumeric		59465-5
619068	Additional Results	Alphanumeric		82939-0
619069	Resources	Alphanumeric		99622-3
619070	Additional Information	Alphanumeric		48767-8
619071	Method	Alphanumeric		85069-3
619072	Genes Analyzed	Alphanumeric		82939-0
619073	Disclaimer	Alphanumeric		62364-5
619074	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81405

81479

81479 (if appropriate for government payers)

Reference Values:

An interpretive report will be provided.