MAYO CLINIC LABORATORIES

# Test Definition: NENZ

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

**Reporting Title:** RBC Enzyme 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 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 panel aids in the diagnosis and genetic counseling of individuals with inherited red blood cell enzymopathies, possible carrier states, or compound variants with severity modulating interactions. This test is best interpreted in the context of protein functional findings by enzymatic assay, complete blood cell count, and peripheral blood findings. This complete interpretation can be provided by also ordering the EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood or HAEV1 / Hemolytic Anemia 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 a more precise interpretation of results.

## Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

## Necessary Information:

1. **Metabolic Hematology Next-Generation Sequencing (NGS) Patient Information** **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 previous enzyme testing, 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:**
1. 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

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## Forms:

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 | Type | Unit | LOINC® |
|---|---|---|---|---|
| 619048 | Test Description | Alphanumeric | | 62364-5 |
| 619049 | Specimen | Alphanumeric | | 31208-2 |
| 619050 | Source | Alphanumeric | | 31208-2 |
| 619051 | Result Summary | Alphanumeric | | 50397-9 |
| 619052 | Result | Alphanumeric | | 82939-0 |
| 619053 | Interpretation | Alphanumeric | | 59465-5 |
| 619054 | Additional Results | Alphanumeric | | 82939-0 |
| 619055 | Resources | Alphanumeric | | 99622-3 |
| 619056 | Additional Information | Alphanumeric | | 48767-8 |
| 619057 | Method | Alphanumeric | | 85069-3 |
| 619058 | Genes Analyzed | Alphanumeric | | 82939-0 |
| 619059 | Disclaimer | Alphanumeric | | 62364-5 |
| 619060 | 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.