
Reporting Title: RBC Enzyme Sequencing, NGS**Performing Location:** Mayo Clinic Laboratories - Rochester Main Campus**Ordering Guidance:**

Multiple hematology gene panels are available. For more information see [Hereditary Hemolytic Anemia Gene Panel and Subpanel Comparison](#).

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

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.