
Reporting Title: CYB5 and CYB5 Reductase, NGS**Performing Location:** Rochester**Ordering Guidance:**

This test should be performed after more common causes of methemoglobinemia have been eliminated. To assess for more common causes of methemoglobinemia, order MEV1 / Methemoglobinemia Evaluation, Blood.

Testing for the *CYB5A* and *CYB5R3* genes as part of a customized panel is available. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

[Multiple gene panels are available. For more information see NHEP and Subpanel Comparison Gene List.](#)

Targeted testing for familial variants (also called site-specific or known variants testing) is available for the *CYB5A* and *CYB5R3* genes. 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 functional findings by enzymatic assay and complete blood cell count analysis. This complete interpretation can be provided by also ordering the MEV1 / Methemoglobinemia Evaluation, Blood. Fill out the information sheet and indicate that a next-generation sequencing test was also ordered. Providing complete blood cell count data and clinical notes will also allow 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 clinical presentation and/or laboratory findings.

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.**

Test Definition: NCYB

Recessive Congenital Methemoglobinemia,
CYB5 and CYB5 Reductase Genetic Analysis,
Next-Generation Sequencing, Varies

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®
618978	Test Description	Alphanumeric		62364-5
618979	Specimen	Alphanumeric		31208-2
618980	Source	Alphanumeric		31208-2
618981	Result Summary	Alphanumeric		50397-9
618982	Result	Alphanumeric		82939-0
618983	Interpretation	Alphanumeric		59465-5
618984	Additional Results	Alphanumeric		82939-0
618985	Resources	Alphanumeric		99622-3
618986	Additional Information	Alphanumeric		48767-8
618987	Method	Alphanumeric		85069-3
618988	Genes Analyzed	Alphanumeric		82939-0
618989	Disclaimer	Alphanumeric		62364-5
618990	Released By	Alphanumeric		18771-6

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

Supplemental Report:

Supplemental

CPT Code Information:

81479

Reference Values:

An interpretive report will be provided.