
Reporting Title: CYB5 and CYB5 Reductase, NGS**Performing Location:** Mayo Clinic Laboratories - Rochester Main Campus**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.

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

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:**Necessary Information:**

- [Metabolic Hematology Next-Generation Sequencing \(NGS\) Patient Information](#) is strongly recommended but not required. Testing may proceed without the patient information; however, it aids in providing a more thorough interpretation. Ordering healthcare professionals are strongly encouraged to complete the form and send it with the specimen
- 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 any previous bone marrow studies, clinical or morphologic presentation.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a bone marrow transplant, call 800-533-1710.

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD)

Specimen Volume: 3 mL

Collection Instructions:

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

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

Test Definition: NCYB

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

Additional Information: 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.

Forms:

- 1. 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 \(Spanish\) \(T826\)](#)
- [Metabolic Hematology Next-Generation Sequencing \(NGS\) Patient Information \(T816\)](#)
- 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.

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