Test Definition: GNMY9

MYH9-Related Disorders, MYH9 Gene, Next-Generation Sequencing, Varies

Reporting Title: MYH9 Gene, Full Gene NGS
Performing Location: Mayo Clinic Laboratories - Rochester Main Campus

Ordering Guidance:

This test is designed to evaluate *MYH9*-related disorders, including May-Hegglin disorder/anomaly and Sebastian syndrome, and to be utilized for genetic confirmation of a phenotypic diagnosis of an *MYH9*-related disorder.

If testing for hereditary platelet disorders using a larger panel is desired, a 70-gene platelet disorder panel is available; order GNPLT / Platelet Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies.

This test is not designed to evaluate for hereditary bleeding disorders. For patients with clinical suspicion of an inherited bleeding disorder, it is important to exclude plasmatic factor deficiencies (eg, von Willebrand disease, hemophilia, or other factor deficiencies) prior to considering an inherited platelet function defect. If bleeding is the indication for testing and testing for hereditary bleeding disorders is desired, bleeding panels are available. For more information see GNBLF / Bleeding Disorders, Focused Gene Panel, Next-Generation Sequencing, Varies or GNBLC / Bleeding Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies.

For assessment of hereditary platelet disorders that have ultrastructural abnormalities, such as gray platelet syndrome, order PTEM / Platelet Transmission Electron Microscopic Study, Whole Blood.

For assessment of hereditary platelet disorders due to quantitative surface glycoprotein deficiencies, order PLAFL / Platelet Glycoprotein Flow Platelet Surface Glycoprotein by Flow Cytometry, Blood.

Testing for the *MYH9* gene as part of a customized 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 *MYH9* gene. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Necessary Information:

Platelet Esoteric Testing 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.

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)

MAYO CLINIC LABORATORIES

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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. **Platelet Esoteric Testing Patient Information 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 an Coagulation Test Request (T753) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619300	Test Description	Alphanumeric		62364-5
619301	Specimen	Alphanumeric		31208-2
619302	Source	Alphanumeric		31208-2
619303	Result Summary	Alphanumeric		50397-9
619304	Result	Alphanumeric		82939-0
619305	Interpretation	Alphanumeric		69047-9
619306	Additional Results	Alphanumeric		82939-0
619307	Resources	Alphanumeric		99622-3
619308	Additional Information	Alphanumeric		48767-8
619309	Method	Alphanumeric		85069-3
619310	Genes Analyzed	Alphanumeric		82939-0
619311	Disclaimer	Alphanumeric		62364-5
619312	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.