Test Definition: GNSPD

Platelet Storage Pool Deficiency Gene Panel, Next-Generation Sequencing, Varies

Reporting Title: Storage Pool Deficiency Panel, NGS
Performing Location: Mayo Clinic Laboratories - Rochester Main Campus

Ordering Guidance:

This test is designed to evaluate a variety of hereditary platelet storage pool deficiencies and to be utilized for genetic confirmation of a phenotypic diagnosis of a platelet storage pool deficiency. If testing for hereditary platelet disorders using a larger, comprehensive panel is desired, a 70-gene platelet panel is available; order GNPLT / Platelet Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies.

This test is not designed to evaluate 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.

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.

Shipping Instructions:

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 healthcare professionals 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. For information about testing patients who have received a bone marrow transplant, call 800-533-1710..
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.**

MAYO CLINIC LABORATORIES

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Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days
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. **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®
619328	Test Description	Alphanumeric		62364-5
619329	Specimen	Alphanumeric		31208-2
619330	Source	Alphanumeric		31208-2
619331	Result Summary	Alphanumeric		50397-9
619332	Result	Alphanumeric		82939-0
619333	Interpretation	Alphanumeric		69047-9
619334	Additional Results	Alphanumeric		82939-0
619335	Resources	Alphanumeric		99622-3
619336	Additional Information	Alphanumeric		48767-8
619337	Method	Alphanumeric		85069-3
619338	Genes Analyzed	Alphanumeric		82939-0
619339	Disclaimer	Alphanumeric		62364-5
619340	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.