Test Definition: GNPFD

Platelet Function Defect Gene Panel, Next-Generation Sequencing, Varies

Reporting Title: Platelet Function Defect Panel, NGS
Performing Location: Rochester

Ordering Guidance:

This test is designed to evaluate a variety of hereditary platelet function defect disorders and to be utilized for genetic confirmation of a phenotypic diagnosis of a platelet function defect disorder. 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 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. 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.

Customization of this panel and single gene analysis for any gene present on this panel are 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.

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)

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

Specimen Minimum Volume:
1 mL

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®
619356	Test Description	Alphanumeric		62364-5
619357	Specimen	Alphanumeric		31208-2
619358	Source	Alphanumeric		31208-2
619359	Result Summary	Alphanumeric		50397-9
619360	Result	Alphanumeric		82939-0
619361	Interpretation	Alphanumeric		59465-5
619362	Additional Results	Alphanumeric		82939-0
619363	Resources	Alphanumeric		99622-3
619364	Additional Information	Alphanumeric		48767-8
619365	Method	Alphanumeric		85069-3
619366	Genes Analyzed	Alphanumeric		82939-0
619367	Disclaimer	Alphanumeric		62364-5

Result ID	Reporting Name	Type	Unit	LOINC®
619368	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.