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MAYO CLINIC LABORATORIES

Test Definition: GNSPD

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

Overview

Useful For

Evaluating hereditary platelet storage pool deficiencies in patients with a personal or family history suggestive of a hereditary platelet storage pool deficiency

Diagnosing hereditary platelet storage pool deficiencies for patients in whom phenotypic testing is nondiagnostic, but there is a strong clinical suspicion of the hereditary platelet storage pool deficiency

Confirming a hereditary platelet storage pool deficiency diagnosis with the identification of a known or suspected disease-causing alteration in one or more of 24 genes associated with a variety of hereditary platelet storage pool deficiencies

Determining the disease-causing alterations within one or more of these 24 genes to delineate the underlying molecular defect in a patient with a laboratory diagnosis of a platelet storage pool deficiency

Identifying the causative alteration for genetic counseling purposes

Prognosis and risk assessment based on genotype-phenotype correlations

Providing a prognosis in syndromic hereditary platelet storage pool deficiencies

Carrier testing for close family members of an individual with a hereditary platelet storage pool deficiency diagnosis

This test is **not intended for** prenatal diagnosis

Genetics Test Information

This test utilizes next-generation sequencing to detect single nucleotide and copy number variants in 24 genes associated with a variety of hereditary platelet storage pool deficiency disorders: *ABCC4*, *AP3B1*, *AP3D1*, *BLOC1S3*, *BLOC1S5*, *BLOC1S6*, *DTNBP1*, *FLI1*, *GFI1B*, *HPS1*, *HPS3*, *HPS4*, *HPS5*, *HPS6*, *LYST*, *NBEA*, *NBEAL2*, *ORAI1*, *PLAU*, *STIM1*, *STXBP2*, *VIPAS39*, *VPS33B*, and *WAS*. See Targeted Genes and Methodology Details for Platelet Storage Pool Deficiency Gene Panel and Method Description for additional details.

Identification of a disease-causing variant may assist with diagnosis, prognosis, clinical management, recurrence risk assessment, familial screening, and genetic counseling for a variety of hereditary platelet storage pool deficiency disorders.

Testing Algorithm

The clinical workup for detecting inherited platelet disorders should begin with a careful review of complete blood cell count and peripheral blood smear results, as well as other platelet tests, such as light transmission platelet aggregometry, electrical impedance whole blood aggregometry, platelet function analyzer 100 (PFA-100), platelet transmission electron microscopy (TEM), and platelet flow cytometric analysis. TEM is an essential tool for laboratory

diagnosis of various hereditary platelet disorders that have ultrastructural abnormalities, such as gray platelet syndrome. Flow cytometry is the preferred method to assess hereditary platelet disorders due to quantitative surface glycoprotein deficiencies.

Platelet laboratory testing may not be able to identify all inherited platelet disorders. Occasionally, the clinical picture may be consistent with a defect in primary hemostasis, but the results of platelet function tests may be normal or non-diagnostic.

Genetic testing for hereditary platelet disorders is indicated if:
-Platelet tests indicate a deficiency or functional abnormality
-There is a clinical suspicion for a hereditary platelet disorder due to family history or patient's clinical presentation
-Acquired causes of deficiencies associated with platelet disorders have been excluded

If a platelet disorder is a concern, a set of clinical guidelines from the British Society for Haematology on testing for heritable platelet disorders is freely available.

Special Instructions

- Informed Consent for Genetic Testing
- Platelet Esoteric Testing Patient Information
- Informed Consent for Genetic Testing (Spanish)
- Targeted Genes and Methodology Details for Platelet Storage Pool Deficiency Gene Panel

Method Name

Sequence Capture and Targeted Next-Generation Sequencing (NGS) followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing

NY State Available

Yes

Specimen

Specimen Type

Varies

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.

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 Required

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.**
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 Minimum Volume

1 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical & Interpretive

Clinical Information

Platelets have essential roles in primary hemostasis. Patients with either hereditary or acquired platelet disorders usually have bleeding diathesis, which can potentially be life threatening. They may also have issues with the development and/or functioning of major organs.(2) Inherited platelet disorders can be syndromic (ie, associated with current or future development of other organ system defects) or nonsyndromic (ie, isolated to thrombocytopenia with no other organ system defects).

A reliable laboratory diagnosis of a platelet disorder can significantly impact patients' and, potentially, their family members' clinical management and outcome. Identification of an alteration that is known or suspected to cause disease aids in confirmation of the diagnosis and potentially provides prognostic information, especially in syndromic inherited platelet disorders.

This panel evaluates 24 genes associated with a variety of hereditary platelet storage pool deficiencies, including reduced adenosine diphosphate (ADP)-induced platelet aggregation; Hermansky-Pudlak syndrome; Paris-Trousseau-Jacobsen syndrome; platelet-type bleeding disorder 17; Chediak-Higashi syndrome; autism with platelet dense granule defect; gray platelet syndrome; autosomal dominant tubular aggregate myopathy-2; Quebec platelet disorder; Stormorken syndrome; York platelet syndrome; familial hemophagocytic lymphohistiocytosis type 5; ARC syndromes (arthrogryposis, renal dysfunction, and cholestasis) 1 and 2; and Wiskott-Aldrich syndrome.

The risk of developing bleeding or other phenotypic features associated with these disorders and syndromes varies. Several of the genes on this panel have established bleeding, thrombocytopenia, or other hematologic or nonhematologic disease associations. Several of the genes on this panel also have expert group guidelines.(1,3-5)

It is recommended that genetic testing be offered to all patients suspected of having a heritable platelet disorder since some patients may have normal platelet laboratory testing results.(1,6)

Reference Values

An interpretive report will be provided.

Interpretation

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations.(7) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Cautions

Clinical Correlations:
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

If testing was performed because of a clinically significant family history, it is often useful to first test an affected family member. Detection of a reportable variant in an affected family member would allow for more informative testing of at-risk individuals.

To discuss the availability of additional testing options or for assistance in the interpretation of these results, contact the Mayo Clinic Laboratories genetic counselors at 800-533-1710.

Technical Limitations:
Next-generation sequencing may not detect all types of genomic variants. In rare cases, false-negative or false-positive results may occur. The depth of coverage may be variable for some target regions; assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. Confirmation of select reportable variants will be performed by alternate methodologies based on internal laboratory criteria.

This test is validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47 bp. Deletions-insertions (delins) of 40 or more bp, including mobile element insertions, may be less reliably detected than smaller delins.

This analysis targets single and multi-exon deletions/duplications; however, in some instances, single exon resolution cannot be achieved due to isolated reduction in sequence coverage or inherent genomic complexity. Balanced structural rearrangements (such as translocations and inversions) may not be detected.

Deletion/duplication events that extend past the genes included on the panel may occur. In these instances, genes included in the ordered test are provided on the report and interpreted, and genomic breakpoints are reported if they are confirmed. However, copy number variants for genes not listed in the Method Description are typically not reported or interpreted for haploinsufficiency/triplosensitivity. CMACB / Chromosomal Microarray, Congenital, Blood; WESPR / Panel to Whole Exome Sequencing Reflex Test, Varies; or WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies is recommended for a full interpretation of deletions/duplications predicted to extend past the genes included on the panel.

This test is not designed to detect low levels of mosaicism or differentiate between somatic mutations and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

For detailed information regarding gene specific performance and technical limitations, see Method Description or contact a laboratory genetic counselor.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reclassification of Variants:
Currently, it is not standard practice for the laboratory to systematically review previously classified variants on a regular basis. The laboratory encourages healthcare professionals to contact the laboratory at any time to learn how the classification of a particular variant may have changed over time. Due to broadening genetic knowledge, it is possible that the laboratory may discover new information of relevance to the patient. Should that occur, the laboratory may issue an amended report.

Variant Evaluation:
Evaluation and categorization of variants are performed using published American College of Medical Genetics and Genomics and the Association for Molecular Pathology recommendations as a guideline.(7) Other gene-specific guidelines may also be considered. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. Variants classified as benign or likely benign are not reported.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and periodic updates to these tools may cause predictions to change over time. Results from in silico evaluation tools should be interpreted with caution and professional clinical judgment.

Rarely, incidental or secondary findings may implicate another predisposition or presence of active disease. These findings will be carefully reviewed to determine whether they will be reported.

Clinical Reference

1. Gomez K, Anderson J, Baker P, et al. Clinical and laboratory diagnosis of heritable platelet disorders in adults and children: a British Society for Haematology Guideline. Brit J Haematol. 2021;195(1):46-72
2. Nurden AT, Freson K, Selifsohn U. Inherited platelet disorders. Haemophilia. 2012;18 Suppl 4:154-160
3. International Society on Thrombosis and Haemostasis: Bleeding Thrombotic and Platelet Disorder TIER1 genes. ISTH; 2018. Updated July 26, 2024. Accessed March 2, 2026. Available at: www.isth.org/page/GinTh_GeneLists
4. Megy K, Downes K, Simeoni I, et al. Curated disease-causing genes for bleeding, thrombotic, and platelet disorders: Communication from the SSC of the ISTH. J Thromb Haemost. 2019;17(8):1253-1260
5. Bolton-Maggs PHB, Chalmers EA, Collins PW, et al. A review of inherited platelet disorders with guidelines for their management on behalf of the UKHCDO. Brit J Haematol. 2006;135(5):603-633
6. Watson SP, Lowe GC, Lordkipanidze M, Morgan NV. GAPP consortium: Genotyping and phenotyping of platelet function disorders. J Thromb Haemost. 2013;11 Suppl 1:351-363
7. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015;17(5):405-424

Performance

Method Description

Next-generation sequencing (NGS) and/or Sanger sequencing are performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed, as well as some other regions that have known disease-causing variants. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth over 30X. Sensitivity is estimated at above 99% for single nucleotide variants, above 94% for deletion-insertions less than 40 base pairs (bp), and above 95% for deletions up to 75 bp and insertions up to 47 bp. NGS and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. See Targeted Genes and Methodology Details for Platelet Storage Pool Deficiency Gene Panel and Methodology Details for details regarding the targeted genes analyzed for each test and specific gene regions not routinely covered.(Unpublished Mayo method)

Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genes analyzed: *ABCC4*, *AP3B1*, *AP3D1*, *BLOC1S3*, *BLOC1S5*, *BLOC1S6*, *DTNBP1*, *FLI1*, *GFI1B*, *HPS1*, *HPS3*, *HPS4*, *HPS5*, *HPS6*, *LYST*, *NBEA*, *NBEAL2*, *ORAI1*, *PLAU*, *STIM1*, *STXBP2*, *VIPAS39*, *VPS33B*, and *WAS*

PDF Report

No

Day(s) Performed

Varies

Report Available

28 to 42 days

Specimen Retention Time

Whole blood: 2 weeks (if available); Extracted DNA: 3 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81443

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
GNSPD	Storage Pool Deficiency Panel, NGS	105335-4

Result ID	Test Result Name	Result LOINC® Value
619328	Test Description	62364-5
619329	Specimen	31208-2
619330	Source	31208-2
619331	Result Summary	50397-9
619332	Result	82939-0
619333	Interpretation	69047-9
619334	Additional Results	82939-0
619335	Resources	99622-3
619336	Additional Information	48767-8
619337	Method	85069-3
619338	Genes Analyzed	82939-0
619339	Disclaimer	62364-5
619340	Released By	18771-6