Lab Dept: Anatomic Pathology

Test Name: INHERITED THROMBOCYTOPENIA PANEL

General Information

Lab Order Codes: ITPA

Synonyms: Thrombocytopenia Panel

CPT Codes: 81404 – Molecular pathology, Level 5
81406 – Molecular pathology, Level 6
81479 – Unlisted molecular pathology

Test Includes: Detection of germline variants in 23 genes known to cause inherited thrombocytopenia.

Logistics

Test Indications: Clarification and/or confirmation of diagnosis in a patient with clinical findings of thrombocytopenia or an associated genetic syndrome when patien's history suggests the possibility of multiple inherited thrombocytopenia disorders.

Identification of carriers with family history of an unspecified thrombocytopenia disorder to provide accurate reproductive risk assessment.

Inherited thrombocytopenia (ITP) is a heterogeneous group of disorders characterized by low platelet counts typically less than 150,000/uL, but often can vary with age, gender and ethnic background. Symptoms may include: purpura, petechiae, prolonged bleeding from cuts, epistaxis, gum bleeding, excessive bleeding after surgery, hemoptysis, hematuria and menorrhagia in women. Severe inherited thrombocytopenia can present in the newborn period, while mild thrombocytopenia may remain undiagnosed until incidental detection on routine blood testing in adulthood. Some inherited types of thrombocytopenia have only hematologic manifestations, such as differences in platelet size or distinctive granulocyte inclusions, while other syndromic types present with additional non-hematologic manifestations. Certain types of inherited thrombocytopenia cuase predisposition to acute myelogenous leukemia or myelodysplastic syndromes.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Blood Centers of Wisconsin (BCW test: 4840)

Phone Numbers: MIN Lab: 612-813-6280

	STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	21 days
Special Instructions:	Please complete all pages of the requisition form. Include Clinical history (including patient's ethnicity, clinical diagnosis, family history and relevant laboratory findings) which is necessary for optimal interpretation of genetic test results and recommendations.

Specimen

Specimen Type:	Whole blood
Container:	Lavender (EDTA) tube
Draw Volume:	3-5 mL EDTA whole blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection, mix specimen gently after collection
Special Processing:	Lab Staff: Do Not centrifuge. Send in original collection container. Ship overnight per instructions on requisition form.
Patient Preparation:	None
Sample Rejection:	Clotted sample, mislabeled or unlabeled specimen

Interpretive

Reference Range:	An interpretive report will be provided
Critical Values:	N/A
Limitations:	The analytical sensitivity of this test is >99% for single nucleotide changes and insertions and deletions of less than 20 bp. This analysis does not detect large deletions, duplications or variants that are outside the regions sequenced.
Methodology:	Capture Hybridization and Next Generation Sequencing (MiSeq)
References:	Blood Centers of Wisconsin (January 2019)