Test Definition: GNADM

Hereditary Thrombotic Thrombocytopenic Purpura, ADAMTS13 Gene, Next-Generation Sequencing, Varies

Reporting Title: ADAMTS13 Gene, Full Gene NGS
Performing Location: Rochester

Ordering Guidance:

This test is designed to detect disease-causing variants in the ADAMTS13 gene and to be utilized for genetic confirmation of a clinical diagnosis of hereditary thrombotic thrombocytopenic purpura (TTP). Genetic testing for hereditary TTP should only be considered if a patient's clinical presentation and initial ADAMTS-13 activity and functional inhibitor screens indicate a diagnosis.

This test does not measure ADAMTS-13 activity or the presence/absence of inhibitors. For assessment of ADAMTS-13 activity and inhibitor status, order ADM13 / ADAMTS13 Activity and Inhibitor Profile, Plasma.

Testing for the ADAMTS13 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 ADAMTS13 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:

Rare Coagulation Disorder 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 it 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. Rare Coagulation Disorder Patient Information (T824) 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)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619230	Test Description	Alphanumeric		62364-5
619231	Specimen	Alphanumeric		31208-2
619232	Source	Alphanumeric		31208-2
619233	Result Summary	Alphanumeric		50397-9
619234	Result	Alphanumeric		82939-0
619235	Interpretation	Alphanumeric		69047-9
619236	Additional Results	Alphanumeric		82939-0
619237	Resources	Alphanumeric		99622-3
619238	Additional Information	Alphanumeric		48767-8
619239	Method	Alphanumeric		85069-3
619240	Genes Analyzed	Alphanumeric		82939-0
619241	Disclaimer	Alphanumeric		62364-5
619242	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.