MAYO CLINIC LABORATORIES

Test Definition: GNANT

Antithrombin Deficiency, SERPINC1 Gene, Next-Generation Sequencing, Varies

Reporting Title: SERPINC1 Gene, Full Gene NGS
Performing Location: Rochester

Ordering Guidance:

This test should only be considered if clinical and family history, initial coagulation screens, and/or initial antithrombin (AT) activity and antigen testing results suggest a diagnosis of antithrombin deficiency (see Testing Algorithm).

This test does not measure AT activity levels. For assessment of AT activity, order ATTF / Antithrombin Activity, Plasma.

If genetic testing for hereditary blood clotting disorders using a larger panel is desired, a 16-gene comprehensive thrombosis panel is available; order GNTHR / Thrombosis Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies.

Testing for the *SERPINC1* 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 *SERPINC1* 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 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

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)
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®
619006	Test Description	Alphanumeric		62364-5
619007	Specimen	Alphanumeric		31208-2
619008	Source	Alphanumeric		31208-2
619009	Result Summary	Alphanumeric		50397-9
619011	Interpretation	Alphanumeric		59465-5
619012	Additional Results	Alphanumeric		82939-0
619013	Resources	Alphanumeric		99622-3
619014	Additional Information	Alphanumeric		48767-8
619015	Method	Alphanumeric		85069-3
619016	Genes Analyzed	Alphanumeric		82939-0
619017	Disclaimer	Alphanumeric		62364-5
619018	Released By	Alphanumeric		18771-6
619010	Result	Alphanumeric		82939-0

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.