Test Definition: GNANG

Hereditary Angioedema Focused Gene Panel, Next-Generation Sequencing, Varies

Reporting Title: Hereditary Angioedema Panel, NGS
Performing Location: Mayo Clinic Laboratories - Rochester Main Campus

Ordering Guidance:

This test is designed to detect disease-causing variants in the *F12, KNG1*, and *PLG* genes and to be utilized for genetic confirmation of a clinical diagnosis of hereditary angioedema with normal C1 inhibitor (HAE with normal C1INH) or factor XII deficiency.

Genetic testing for HAE with normal C1INH should only be considered if there is a documented family history of angioedema that does not respond to chronic, high-dose antihistamine therapy, normal complement studies, normal C1-INH level and function, and no exposure to medications that could cause angioedema, such as angiotensin-converting enzyme inhibitors or nonsteroidal anti-inflammatory drugs.

Genetic testing for factor XII deficiency should only be considered if clinical and family history, initial coagulation screens, or initial activity tests indicate a diagnosis.

This test does not measure complement 4, C1INH antigen, C1INH functional, or factor XII activity levels.
-For assessment of C4, order C4 / Complement C4, Serum.
-For assessment of C1INH antigen, order C1ES / C1 Esterase Inhibitor Antigen, Serum.
-For assessment of functional C1INH , order C1INF / C1 Esterase Inhibitor, Functional, Serum.
-For assessment of factor XII activity, order F_12 / Coagulation Factor XII Activity Assay, Plasma.

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:

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

MAYO CLINIC LABORATORIES

Test Definition: GNANG

Hereditary Angioedema Focused Gene Panel, Next-Generation Sequencing, Varies

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®
619216	Test Description	Alphanumeric		62364-5
619217	Specimen	Alphanumeric		31208-2
619218	Source	Alphanumeric		31208-2
619219	Result Summary	Alphanumeric		50397-9
619220	Result	Alphanumeric		82939-0
619221	Interpretation	Alphanumeric		59465-5
619222	Additional Results	Alphanumeric		82939-0
619223	Resources	Alphanumeric		99622-3
619224	Additional Information	Alphanumeric		48767-8
619225	Method	Alphanumeric		85069-3
619226	Genes Analyzed	Alphanumeric		82939-0
619227	Disclaimer	Alphanumeric		62364-5
619228	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81479

Reference Values:

Test Definition: GNANG

Hereditary Angioedema Focused Gene Panel,
Next-Generation Sequencing, Varies

An interpretive report will be provided.