MAYO CLINIC LABORATORIES

Test Definition: GNBLC

Bleeding Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies

Reporting Title: Bleeding Comprehensive Panel, NGS
Performing Location: Rochester

Ordering Guidance:

Special coagulation testing for evaluating patients with bleeding or hypocoagulability states should be performed prior to genetic testing. For more information see ALBLD / Bleeding Diathesis Profile, Limited, Plasma.

This test is designed to evaluate a variety of clotting factor-related hereditary bleeding disorders.

If testing for hereditary bleeding disorders using a smaller panel is desired, a six-gene bleeding panel is available; order GNBLF / Bleeding Disorders, Focused Gene Panel, Next-Generation Sequencing, Varies

This test is not designed to evaluate for a single common hereditary bleeding disorder, such as when an individual has a known family history of hemophilia A or B or von Willebrand disease, specifically. If testing for a particular common hereditary bleeding disorder is desired, single gene tests are available for the *F8*, *F9*, and *VWF* genes. See GNHMA / Hemophilia A, *F8* Gene, Next-Generation Sequencing, Varies; GNHMB / Hemophilia B, *F9* Gene, Next-Generation Sequencing, Varies; or GNVWD / von Willebrand Disease, *VWF* and *GP1BA* Genes, Next-Generation Sequencing, Varies.

This test does not evaluate for the presence of inversions in the *F8* gene that can cause hemophilia A. If testing for possible inversions in the *F8* gene is desired, order F8INV / Hemophilia A *F8* Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood

This test is not designed to evaluate for hereditary thrombosis disorders. If thrombosis is the indication for testing and testing for hereditary thrombosis disorders is desired, order GNTHR / Thrombosis Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies

This test is not designed to evaluate for inherited platelet disorders. If a platelet disorder is suspected and comprehensive testing for platelet disorders is desired, order GNPLT / Platelet Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies

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 mutations 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.

Additional Testing Requirements:

All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen as this must be a different order number than the prenatal specimen.

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:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
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

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient
Additional information:
1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid.
2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated
Additional Information:
1. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.
2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Acceptable:

Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 Flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated
Additional Information:
All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

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®
619258	Test Description	Alphanumeric		62364-5
619259	Specimen	Alphanumeric		31208-2
619260	Source	Alphanumeric		31208-2
619261	Result Summary	Alphanumeric		50397-9
619262	Result	Alphanumeric		82939-0
619263	Interpretation	Alphanumeric		59465-5
619264	Additional Results	Alphanumeric		82939-0
619265	Resources	Alphanumeric		99622-3
619266	Additional Information	Alphanumeric		48767-8
619267	Method	Alphanumeric		85069-3
619268	Genes Analyzed	Alphanumeric		82939-0
619269	Disclaimer	Alphanumeric		62364-5
619270	Released By	Alphanumeric		18771-6

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

Supplemental Report:

Supplemental

CPT Code Information:

81443
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
88240-Cryopreservation (if appropriate)
88235-Amniotic fluid culture (if appropriate)
81265-Maternal cell contamination (if appropriate)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
_STR1	Comp Analysis using STR (Bill only)	1	81265	No	No, (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)	1	81266	No	No, (Bill only)
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test	1	88235	No	Yes
MATCC	Maternal Cell Contamination, B	1	81265	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	Released By	Alphanumeric		18771-6
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6
MATCC	53285	Result Summary	Alphanumeric		50397-9
MATCC	53286	Result	Alphanumeric		40704-9
MATCC	53287	Interpretation	Alphanumeric		69047-9
MATCC	53288	Reason for referral	Alphanumeric		42349-1
MATCC	53289	Specimen	Alphanumeric		31208-2
MATCC	53290	Source	Alphanumeric		31208-2

MATCC	53291	Released By	Alphanumeric		18771-6
MATCC	55150	Method	Alphanumeric		85069-3

Reference Values:

An interpretive report will be provided.