
Reporting Title: F13A1 and B Genes, Full Gene NGS**Performing Location:** Rochester**Ordering Guidance:**

Special coagulation testing for factor XIII (FXIII) activity should be performed prior to any genetic testing. For assessment of FXIII activity, order ALBLD / Bleeding Diathesis Profile, Limited, Plasma, which includes the factor XIII screening assay.

Genetic testing should only be considered if clinical and family history, initial coagulation screens, or initial activity tests indicate a diagnosis of FXIII deficiency (see Testing Algorithm).

If genetic testing for hereditary bleeding disorders using a larger panel is desired, both a 6-gene focused bleeding panel and a 25-gene comprehensive bleeding panel are available. For more information see GNBLF / Bleeding Disorders, Focused Gene Panel, Next-Generation Sequencing, Varies or GNBLC / Bleeding Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies.

Testing for the F13A1 and F13B genes 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 mutations testing) is available for the F13A1 and F13B genes. 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

Prenatal Specimens

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.
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.

Specimen Minimum Volume:

Blood: 1 mL; Amniotic fluid: 10 mL; Other specimen types: See Specimen Required

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		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No
CULAF	CG767	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619146	Test Description	Alphanumeric		62364-5
619147	Specimen	Alphanumeric		31208-2
619148	Source	Alphanumeric		31208-2
619149	Result Summary	Alphanumeric		50397-9
619150	Result	Alphanumeric		82939-0
619151	Interpretation	Alphanumeric		69047-9
619152	Additional Results	Alphanumeric		82939-0
619153	Resources	Alphanumeric		99622-3
619154	Additional Information	Alphanumeric		48767-8
619155	Method	Alphanumeric		85069-3
619156	Genes Analyzed	Alphanumeric		82939-0
619157	Disclaimer	Alphanumeric		62364-5
619158	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81479
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	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes
MATCC	Maternal Cell Contamination, B			No	Yes
_STR1	Comp Analysis using STR (Bill only)			No	No (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)			No	No (Bill only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
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
CULAF	52304	Result Summary	Alphanumeric		50397-9



Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
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
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	55150	Method	Alphanumeric		85069-3
MATCC	53291	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided.