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**Reporting Title:** FGA/B/G Genes, Full Gene NGS**Performing Location:** Rochester**Ordering Guidance:**

This test is designed to detect single nucleotide and copy number variants in the FGA, FGB, and FGG genes associated with congenital fibrinogen disorders (CFD).

This test should only be considered if coagulation screening tests measuring thrombin clotting time (TT; with or without reptilase time), clottable fibrinogen, and fibrinogen antigen suggest a quantitative or functional defect in fibrinogen, especially if these findings are similar between family members.

For assessment of thrombin clotting time, order TTSC / Thrombin Time (Bovine), Plasma.

For assessment of fibrinogen function, order FIBTP / Fibrinogen, Plasma.

For assessment of fibrinogen quantity, order FIBAG / Fibrinogen Antigen, Plasma.

If genetic testing for CFD using a larger panel is desired, both a 25-gene comprehensive bleeding panel and a 16-gene comprehensive thrombosis panel are available. See GNBLC / Bleeding Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies; and GNTHR / Thrombosis 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 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.

**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.

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**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.
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®
619160	Test Description	Alphanumeric		62364-5
619161	Specimen	Alphanumeric		31208-2
619162	Source	Alphanumeric		31208-2
619163	Result Summary	Alphanumeric		50397-9
619164	Result	Alphanumeric		82939-0
619165	Interpretation	Alphanumeric		59465-5
619166	Additional Results	Alphanumeric		82939-0
619167	Resources	Alphanumeric		99622-3
619168	Additional Information	Alphanumeric		48767-8
619169	Method	Alphanumeric		85069-3
619170	Genes Analyzed	Alphanumeric		82939-0

Result ID	Reporting Name	Type	Unit	LOINC®
619171	Disclaimer	Alphanumeric		62364-5
619172	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

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
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
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.