

Factor VII Deficiency, F7 Gene, Next-Generation Sequencing, Varies

Reporting Title: F7 Gene, Full Gene NGS

Performing Location: Rochester

Ordering Guidance:

Special coagulation testing for factor VII (FVII) activity should be performed prior to any genetic testing. For assessment of FVII activity, order F_7 / Coagulation Factor VII Activity Assay, Plasma.

This test should only be considered if clinical and family history, initial coagulation screens, and/or initial activity tests indicate a diagnosis of FVII 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 F7 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 mutations testing) is available for the F7 gene. 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



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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:

A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid.
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:

Whole 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

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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 | Туре | 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 | Туре | Unit | LOINC® |
|-----------|------------------------|--------------|------|---------|
| 619090 | Test Description | Alphanumeric | | 62364-5 |
| 619091 | Specimen | Alphanumeric | | 31208-2 |
| 619092 | Source | Alphanumeric | | 31208-2 |
| 619093 | Result Summary | Alphanumeric | | 50397-9 |
| 619094 | Result | Alphanumeric | | 82939-0 |
| 619095 | Interpretation | Alphanumeric | | 59465-5 |
| 619096 | Additional Results | Alphanumeric | | 82939-0 |
| 619097 | Resources | Alphanumeric | | 99622-3 |
| 619098 | Additional Information | Alphanumeric | | 48767-8 |
| 619099 | Method | Alphanumeric | | 85069-3 |
| 619100 | Genes Analyzed | Alphanumeric | | 82939-0 |
| 619101 | Disclaimer | Alphanumeric | | 62364-5 |
| 619102 | Released By | Alphanumeric | | 18771-6 |

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

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Supplemental

CPT Code Information:

81479 88233-Tissue culture, skin, solid tissue biopsy (if appropriate) 88240-Cryopreservation (if appropriate) 88235-Amniotic fluid culture (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 | Туре | 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 |
| CULAF | 52306 | Interpretation | Alphanumeric | | 69965-2 |



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| Test ID | Result ID | Reporting Name | Туре | Unit | LOINC® |
|---------|-----------|------------------------|--------------|------|---------|
| 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.