Reporting Title: F8 Gene, Full Gene NGS
Performing Location Rochester

Ordering Guidance:

Genetic testing for hemophilia A should only be considered in males if clinical and family history, initial coagulation screens, and FVIII activity (F8A / Coagulation Factor VIII Activity Assay, Plasma) indicate a diagnosis of hemophilia A. Causes of acquired (non-genetic) hemophilia A should be excluded prior to genetic testing.

Most patients with less than 1% factor VIII activity and severe hemophilia A have large rearrangements in the *F8* gene called inversions that are not detectable by this testing method. Full gene sequencing for a patient with severe hemophilia A should be performed only if genetic testing for these inversions is negative. See F8INV / Hemophilia A *F8* Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood.

Genetic testing for hemophilia A in females should only be considered if a first-degree male relative has been diagnosed with hemophilia A, if there is a maternal family history of hemophilia A and her mother has not been excluded as a carrier, or if the patient has abnormally low FVIII activity (F8A / Coagulation Factor VIII Activity Assay, Plasma).

For females with bleeding symptoms and no known personal or family history of hemophilia A, consider BDIAL / Bleeding Diathesis Profile, Limited or the specific factor assays.

Prenatal genetic testing should NOT be performed without the prior identification of a familial hemophilia alteration because diagnostic prenatal testing requires an invasive procedure that carries a small but real risk of inducing spontaneous abortion.

Additional Testing Requirements:

Due to the complexity of testing non-peripheral blood, consultation with the laboratory is required for all cord blood samples. All cord blood specimens **must be accompanied** by a maternal blood specimen. Order this test on the cord blood specimen (only 1 sample tube required) and order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Shipping Instructions:

Prenatal Specimens: Advise Express Mail or equivalent if not on courier service. Prenatal specimens can be sent Monday through Thursday and **must be received by 3 p.m. CST on Friday** in order to be processed appropriately.
Blood: Ambient and refrigerate specimens **must** arrive within 7 days and frozen specimens must arrive within 14 days. Collect and package specimen as close to shipping time as possible.

Necessary Information:

Hemophilia A Patient Information **(T712) is required,** see Special Instructions. 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:

Submit only 1 of the following specimens:

Specimen Type: Peripheral blood or cord blood
Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD) or light-blue top (sodium citrate)
Specimen Volume: 3 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.
Specimen Stability Information: Ambient (preferred) 7 days/Refrigerated 7 days/Frozen 14 days

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing.

Specimen Type: Amniotic fluid
Supplies: Refrigerate/Ambient Shipping Box, 5 lb (T329)
Container/Tube: Amniotic fluid container
Specimen Volume: 10-20 mL
Collection Instructions:
1. Optimal timing for specimen collection is during 14 to 18 weeks of gestation, but specimens collected at other weeks of gestation are also accepted.
2. Discard the first 2 mL of amniotic fluid.
3. Place the tubes in a Styrofoam container.
4. Fill remaining space with packing material.
5. Unavoidably, about 1% to 2% of mailed-in specimens are not viable.
6. Bloody specimens are undesirable.
7. If the specimen does not grow in culture, you will be notified within 7 days of receipt.
Additional Information:
A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid
Specimen Stability Information: Refrigerated (preferred) <24 hours/Ambient <24 hours

Specimen Type: Chorionic villi
Supplies: CVS Media (RPMI) and Small Dish (T095)
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20-30 mg
Collection Instructions:
1. Collect specimen by the transabdominal or transcervical method.
2. Transfer the chorionic villi specimen to a Petri dish containing transport medium.
3. Using a stereomicroscope and sterile forceps, assess the quality and quantity of the villi and remove any blood clots and maternal decidua.
Additional Information:
A separate culture charge will be assessed under CULFB / Fibroblast Culture for Genetic Testing
Specimen Stability Information: Refrigerated (preferred) <24 hours/Ambient <24 hours

Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 Flasks approximately 90% confluent
Collection Instructions: Submit confluent cultured cells from another laboratory
Additional Information: There will be no culture charge.
Specimen Stability Information: Ambient (preferred) <24 hours/Refrigerated <24 hours

Forms:

1. Hemophilia A Patient Information **(T712) is required,** see Special Instructions.

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 in Special Instructions:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
3. If not ordering electronically, complete, print, and send a Coagulation Test Request (T753) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies (preferred)	0 hours	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC
113044	F8NGS Result	Alphanumeric		50397-9
113038	Alterations Detected	Alphanumeric		82939-0
113036	Interpretation	Alphanumeric		69047-9
113039	Additional Information	Alphanumeric		48767-8
113040	Method	Alphanumeric		85069-3
113041	Disclaimer	Alphanumeric		62364-5
113042	Panel Gene List	Alphanumeric		21673-9
113043	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
No

CPT Code Information:
81407

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
CULAF	Amniotic Fluid Culture/Genetic Test	1	88235	No	Yes
CULFB	Fibroblast Culture for Genetic Test	1	88233	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