

Hemophilia A F8 Gene, Intron 1 Inversion Known Mutation Analysis, Prenatal

Reporting Title: HA F8 Intron 1 Inversion, AF or CVS

Performing Location: Rochester

Additional Testing Requirements:

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

All prenatal specimens must be accompanied by a maternal blood specimen. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Shipping Instructions:

Advise Express Mail or equivalent if not on courier service.

Necessary Information:

<u>Hemophilia A Patient Information</u> (T712) **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:

Results will be reported and telephoned or faxed, if requested.

Submit only 1 of the following specimens:

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 5-10 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. If the culture will be performed in conjunction with chromosome analysis and alpha-fetoprotein, a total of approximately 25 to 30 mL will be needed for the combined studies.

Specimen Stability Information: Ambient (preferred) <24 hours/Refrigerated

Additional Information:

- 1. Place the tubes in a Styrofoam container.
- 2. Fill remaining space with packing material.
- 3. Unavoidably, about 1% to 2% of mailed-in specimens are not viable.
- 4. Bloody specimens are undesirable.
- 5. If the specimen does not grow in culture, you will be notified within 7 days of receipt.
- 6. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid.
- **7. 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

Supplies: CVS Media (RPMI) and Small Dish (T095)

Container/Tube: 15-mL tube containing 15 mL of transport media



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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.

Specimen Stability Information: Refrigerated (preferred) <24 hours/Ambient

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.

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. **Specimen Stability Information:** Ambient (preferred) <24 hours/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. Hemophilia A Patient Information (T712) 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 a Coagulation Test Request (T753) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
35138	HA F8 Int1 KM Reason for Referral	Alphanumeric		42349-1
35004	HA F8 Intron 1 Inversion, AF or CVS	Alphanumeric		82342-7
35005	F81P Interpretation	Alphanumeric		69047-9
35006	HA F8 Int1 KM Reviewed By	Alphanumeric		18771-6

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



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No

CPT Code Information:

81403

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
_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)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	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	nanumeric	



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MATCC	55150	l Method	Alphanumeric	85069-3

Reference Values:

An interpretive report will be provided.