

Test Definition: F81B

Hemophilia A F8 Gene, Intron 1 Inversion Known Mutation, Whole Blood

Reporting Title: HA F8 Intron 1 Inversion KM, B **Performing Location:** Rochester

Ordering Guidance:

If a familial variant has not been identified in a severely affected hemophilia A patient, order F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood.

For evaluation of a patient with bleeding symptoms and no known personal history of a bleeding disorder consider ALBLD / Bleeding Diathesis Profile, Limited, Plasma or the specific factor assays.

Additional Testing Requirements:

Due to the complexity of testing nonperipheral blood, consultation with the laboratory is required for all cord blood samples. Order this test on the cord blood specimen (only 1 specimen tube required) and order MATCC / Maternal Cell Contamination, Molecular Analysis, Blood on the maternal specimen.

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:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD) or blue top (3.2% sodium citrate)

Specimen Volume: 4 mL

Collection Instructions:

- 1. Invert several times to mix blood.
- 2. Send whole blood specimen in original tube. **Do not aliquot.**

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 <u>Coagulation Test Request</u> (T753) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	7 days	
	Frozen	7 days	
	Refrigerated	7 days	



Test Definition: F81B

Hemophilia A F8 Gene, Intron 1 Inversion Known Mutation, Whole Blood

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
35137	HA F8 Int1 KM Reason for Referral	Alphanumeric		42349-1
35001	HA F8 Intron 1 Inversion KM, B	Alphanumeric		81762-7
35002	F81B Interpretation	Alphanumeric		69047-9
35003	HA F8 Int1 KM Reviewed By	Alphanumeric		18771-6

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

Supplemental Report:

No

CPT Code Information:

81403

Reflex Tests:

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