

Test Definition: F8INV

Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood

Reporting Title: HA F8 Intron 1/22 Inversion, B

Performing Location: Mayo Clinic Laboratories - Rochester Main Campus

Ordering Guidance:

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 non-peripheral 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, Varies 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	
	Refrigerated	7 days	
	Frozen	7 days	

Result Codes:

Result ID Reporting Name	Туре	Unit	LOINC®
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35760	HA F8 Int 1/22 Reason for Referral	Alphanumeric	42349-1
35761	HA F8 Intron 1/22 Inversion, B	Alphanumeric	81761-9
35762	F8INV Interpretation	Alphanumeric	69047-9
35763	HA F8 Intron 1/22 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	Туре	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.