MAYO CLINIC LABORATORIES

# Test Definition: F822B

Hemophilia A F8 Gene, Intron 22 Inversion
Known Mutation, Whole Blood

**Reporting Title:** HA F8 Intron 22 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 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, Blood on the maternal specimen.

## Necessary Information:

**Hemophilia A Patient Information** (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 Coagulation Test Request (T753) with the specimen.

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

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## Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|---|---|---|---|---|
| 35139 | HA F8 Int22 KM Reason for Referral | Alphanumeric | | 42349-1 |
| 35007 | HA F8 Intron 22 Inversion KM, B | Alphanumeric | | 91679-1 |
| 35008 | F822B Interpretation | Alphanumeric | | 69047-9 |
| 35009 | HA F8 Int22 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.