Lab Dept:	**Coagulation**
Test Name:	**HEMOPHILIA A MOLECULAR INVERSION**

General Information

Lab Order Codes:	HAMI
Synonyms:	Hemophilia A Factor 8 Intron Inversion 1 and 22 Mutation Analysis
CPT Codes:	81403 – F8, inversion analysis, intron 1 and 22A (Molecular Pathology, Level 4)
Test Includes:	Detects the common inversion mutations within the F8 gene. Approximately 50% of affected males with severe hemophilia A have been shown to have an inversion. It is recommended that the F8 inversion mutation be confirmed in the affected male or obligate carrier female prior to testing at-risk individuals.

Logistics

Test Indications:	First –tier molecular testing for males affected with severe hemophilia A when a mutation has not been identified. Determining hemophilia A carrier status for at-risk females, ie, individuals with a family history of severe hemophilia A.
Lab Testing Section:	Coagulation - Sendouts
Referred to:	Mayo Medical Laboratories (MML Test#: F8INV)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	14 - 21 days, performed Monday - Friday
Special Instructions:	All specimens must include a "Hemophilia A Patient Information Sheet" (Mayo Supply T712 obtain from lab) with information including relevant clinical and family history information in order to provide correct interpretation of test results. If informed consent is required, please submit an "Informed Consent for DNA Testing" (Supply T576-obtain from lab) with the specimen. Note: Each molecular coagulation test requested must have its own tube.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) or Yellow top (ACD) tube Alternate tube: Blue top (Sodium Citrate)
Draw Volume:	3 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture. Gently invert several times to mix. Send specimen to the lab at room temperature immediately.
Special Processing:	Lab Staff: Specimens should remain in original collection containers. Forward specimen(s) and accompanying paperwork to reference laboratory promptly at ambient/room temperature. **Do Not** freeze.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	An interpretive report will be provided.
Critical Values:	N/A
Limitations:	Obtaining a medical genetics or hematology (coagulation) consultation is advisable. Molecular genetic or hemophilia center consultation is available for all possible hemophilia A cases and is particularly indicated in complex cases or in situations in which diagnosis is atypical or uncertain. This assay detects only F8 intron 1 and 22 inversion mutations. Thus, a negative result does not exclude the presence of other mutations in F8. The intron 1 and 22 inversion mutations targeted by this assay are found in approximately 50% of individuals with severe hemophilia A; the assay may be uninformative for a number of families. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete. A previous bone marrow transplant from an allogenic donor will interfere with testing. Contact Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Methodology:	Polymerase Chain Reaction (PCR) or Inverse Shifting-Polymerase Chain Reaction (IS-PCR)
References:	Mayo Medical Laboratories Web Page November 2014 (800) 533-1710
Updates:	2/12/2013: CPT update 11/5/2014: Method update, previously listed as Southern Blot.