Lab Dept: Coagulation

Test Name: HEMOPHILIA A (F8) 2 INVERSIONS REFLEX

General Information

Lab Order Codes: HEMPA

Synonyms: Hemophilia A (Factor 8) 2 Inversions with Reflex to Sequencing and Deletion Duplication

CPT Codes: 81403 – F8, inversion analysis, intron 1 and intron 22A (Molecular Pathology, Level 4)
81406 – F8, deletion/duplication analysis (Molecular Pathology, Level 7)
81407 – F8, full gene sequence

Test Includes: F8 inversion testing is performed on all samples. If inversion testing does not explain the clinical scenario, F8 gene sequencing will be performed at an additional cost. If inversion testing and sequencing do not explain the clinical scenario, deletion/duplication testing will be performed at an additional cost.

Logistics

Test Indications: Severe deficiency of factor VIII clotting activity is associated with spontaneous joint or deep tissue bleeding. Moderate or mild deficiency is associated with prolonged bleeding after tooth extractions, surgery, or injuries and recurrent or delayed wound healing.

Lab Testing Sections: Coagulation - Sendouts

Referred to: ARUP Laboratories, ARUP Test: 2001614

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Within 35 days

Special Instructions: Fill out Patient History form for Hemophilia A or B. Form is found on: http://www.aruplab.com/guides/ug/tests/iconpdf_48.pdf.

Specimen

Specimen Type: Whole blood

Container:	Lavender top (EDTA) tube **Alternate blood tube:** Yellow (ACD Solution A or B) top tube or Pink (K2 EDTA) top tube
Draw Volume:	3 mL (Minimum: 2 mL) blood
Processed Volume:	Forward entire specimen to reference lab
Collection:	Routine venipuncture
Special Processing:	Lab Staff: **Do Not** Centrifuge. Send specimen in original collection container at refrigerated temperature. Forward promptly.
Patient Preparation:	None
Sample Rejection:	Specimen collected in wrong container, mislabeled or unlabeled specimens

Interpretive

Reference Range:	Interpretive report
Limitations:	Rare diagnostic errors can occur due to primer or probe site mutations. Regulatory region and deep intronic mutations, other than the F8 intron 22-A and intron 1 inversions, will not be detected.
Methodology:	**Inversions:** F8 intron 22-A and intron 1 inversions detected by inverse PCR and electrophoresis. **Sequencing:** Bidirectional sequencing of the F8 coding region and intron-exon boundaries. **Deletion/Duplication:** Multiplex ligation-dependent probe amplification (MPLA) to detect large deletions/duplications in the F8 coding region.
References:	ARUP website November 2014
Updates:	2/7/2012: CPT update 2/12/2013: CPT update