TEST CHANGE

Hemophilia A (*F8*) 2 Inversions, Fetal

2001755, F8 INV FE

Specimen Requirements:	
Patient Preparation:	
Collect:	Fetal Cultured Amniocytes or Cultured CVS AND Maternal Whole Blood Specimen: Lavender (EDTA) or yellow (ACD solution A or B).
Specimen Preparation:	Cultured Amniocytes or Cultured CVS: Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80 percent confluence: (Min: one T-25 flask at 80 percent confluence). Backup cultures must be retained at the client's institution until testing is complete. If ARUP receives a sample below the minimum confluence, Cytogenetics Grow and Send (ARUP test code 0040182) will be added on by ARUP, and additional charges will apply. If clients are unable to culture specimens, Cytogenetics Grow and Send should be added to initial order. Maternal Whole Blood Specimen: Transport 2 mL whole blood (Min: 1 mL)
Transport Temperature:	Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells. Maternal Whole Blood Specimen: Room temperature.
Unacceptable Conditions:	
Remarks:	
Stability:	Cultured Amniocytes or Cultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable
Methodology:	Inverse Polymerase Chain Reaction / Electrophoresis
Note:	
CPT Codes:	81403; 81265 Fetal Cell Contamination (FCC)
New York DOH Approval Status:	Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.

Interpretive Data:

Background Information for Hemophilia A (F8) 2 Inversions, Fetal:
Characteristics: Severe deficiency of factor VIII clotting activity leading to spontaneous joint or deep muscle bleeding. Moderate to mild deficiency is associated with prolonged bleeding after tooth extractions, surgery, or injuries and recurrent or delayed wound healing.
Incidence: 1 in 4,000-5,000 live male births worldwide, rare in females.
Inheritance: X-linked recessive. Of simplex cases, 85 percent of mothers are carriers and 10-15 percent of boys have a de novo mutation.
Penetrance: 100 percent in males and 10 percent in females.

Cause: Deleterious F8 gene mutations.
Clinical Sensitivity: 51 percent of mutations causing severe hemophilia A are detected by F8 inversion testing. This assay does not detect F8 mutations associated with mild or moderate hemophilia A in males.
Methodology: Intron 22-A and intron 1 inversions detected by inverse PCR and electrophoresis.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. F8 mutations, other than the F8 intron 22-A and intron 1 inversions, will not be detected.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Reference Interval:

Refer to~~By~~ report

HOTLINE NOTE: There is a prompt change associated with this test. Refer to the Hotline Test Mix for interface build information.