ARUP LABORATORIES | www.aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Hemophilia A (F8) Sequencing

ARUP test code 2001747

F8 FGS Specimen	Whole Blood

Hemophilia A (F8) Sequencing Interp

Negative

TEST PERFORMED - 2001747
TEST DESCRIPTION - Hemophilia A (F8) Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the F8 gene.

INTERPRETATION
No pathogenic variants in the factor 8 (F8) gene were detected through bidirectional sequencing of all coding regions and intron/exon borders. Large gene deletion, duplication or inversion, deep intronic variants, and regulatory region variants are not detected by this assay.

RECOMMENDATIONS
If the patient is suspected to be affected with hemophilia A, targeted testing for the F8 intron 22-A and intron 1 inversions (ARUP test code 2001759) is recommended.Consideration may be given to diagnostic testing for von Willebrand disease, which may also be associated with decreased factor VIII activity. Medical management should rely on family history and clinical findings.

COMMENTS
Reference Sequence: GenBank # NM_000132.3 (F8)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by Weimin Sun, Ph.D.

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Hemophilia A (F8) Sequencing

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: 98 percent of mutations causing mild to moderate hemophilia A and 43 percent of severe hemophilia A mutations are detected by sequencing.
METHODOLOGY: Bidirectional sequencing of the entire F8 coding region and intron-exon boundaries.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations and gene duplications will not be detected in patients of either sex; large deletions will not be detected in females.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
F8 FGS Specimen	19-310-134977	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemophilia A (F8) Sequencing Interp	19-310-134977	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical