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

Physician: Doctor, Example

Patient: Patient, Example

DOB	7/23/1985
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Deletion/Duplication Analysis by MLPA

ARUP test code 3003144

Deletion/Duplication Interpretation: Negative

Deletion/Duplication Analysis by MLPA

RESULT
No pathogenic variants were detected in the F8 gene.

INTERPRETATION
No pathogenic variants were detected in the F8 gene by deletion/duplication analysis. This result reduces the likelihood that this individual is a carrier of hemophilia A. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. Genetic consultation is recommended. Large deletions and duplications account for approximately 6 percent of causative variants in the F8 gene. These results should therefore be correlated with the results of the Hemophilia A (F8) 2 Inversions testing (ARUP accession no: 24-227-121850) and the Hemophilia A (F8) sequencing (ARUP accession no: 24-227-121851) that were ordered concurrently and will be reported under separate cover. For optimal interpretation of this result, determination of the causative familial variant in an affected family member or obligate carrier is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000132.3 (F8)

This result has been reviewed and approved by [redacted]

Deletion/Duplication Gene: F8 DD

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

BACKGROUND INFORMATION: Hemophilia A (F8) Deletion/Duplication

CHARACTERISTICS: Hemophilia A is characterized by deficiency of factor VIII clotting activity. Less than 1 percent factor VIII activity results in severe deficiency associated with spontaneous joint or deep muscle bleeding. Moderate deficiency (1-5 percent activity) and mild deficiency (6-40 percent activity) are associated with prolonged bleeding after tooth extractions, surgery, or injuries, and recurrent or delayed wound healing. Female carriers of hemophilia A may have increased bleeding tendencies.

EPIDEMIOLOGY: 1 in 5,000 live male births worldwide

CAUSE: Pathogenic F8 germline variants

INHERITANCE: X-linked recessive. In the estimated 30 percent of cases that appear to be de novo, the mother is found to be a carrier at least 80 percent of the time.

PENETRANCE: 100 percent in males. Approximately 30 percent of female carriers have factor VIII activity levels of less than 40 percent and are at risk for bleeding symptoms typically consistent with mild hemophilia A.

CLINICAL SENSITIVITY: Approximately 2 percent of variants causing severe hemophilia A and less than 1 percent of mild to moderate hemophilia A variants are detected by deletion/duplication testing.

METHODOLOGY: Multiplex ligation-dependent probe amplification (MLPA) of the F8 gene.

ANALYTICAL SENSITIVITY/SPECIFICITY: 99 percent

LIMITATIONS: A negative result does not exclude a diagnosis of or carrier status for hemophilia A. Diagnostic errors can occur due to rare sequence variations. Breakpoints for large deletions/duplications will not be determined. Single exon deletion/duplications may not be detected based on the breakpoints of the rearrangement. F8 base pair substitutions, small deletions/duplications, deep intronic, and regulatory region variants will not be detected.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Deletion/Duplication Interpretation	24-227-121852	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Deletion/Duplication Gene	24-227-121852	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