

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	3/5/2005	
Sex:	Female	
Patient Identifiers:	01234567890ABCD, 012345	
Visit Number (FIN):	01234567890ABCD	
Collection Date:	01/01/2017 12:34	

Hemophilia A (F8) Sequencing

ARUP test code 3004241				
Hemophilia A (F8) Specimen	Whole Blood			
Hemophilia A (F8) Interp	Negative			
	RESULT No pathogenic variants were detected in the F8 gene.			
	INTERPRETATION No pathogenic variants were detected in the F8 gene. Thus, this individual's risk of being a carrier of hemophilia A is reduced, but not eliminated. Please refer to the background information included in this report for the methodology and limitations of this test.			
	RECOMMENDATIONS Medical screening and management should rely on clinical findings and family history. Genetic consultation is recommended. If this individual is at-risk for being a carrier of hemophilia A, targeted testing for the F8 intron 22-A and intron 1 inversions (ARUP test code 2001759) and F8 deletion/duplication analysis (Deletion/Duplication Analysis by MLPA, ARUP test code 3003144) are recommended. For optimal interpretation of this result, confirmation of the diagnosis and determination of the causative familial variant in an affected family member or obligate carrier is recommended.			
	COMMENTS Likely benign and benign variants are not reported. Variants in the following region(s) may not be detected by NGS with sufficient confidence in this sample due to technical limitations: None			
	This result has been reviewed and approved by			
	BACKGROUND INFORMATION: Hemophilia A (F8) Sequencing			
	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			

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com 500 Chipeta Way, Salt Lake City, UT 84108-1221 Jonathan R. Genzen, MD, PhD, Laboratory Director Patient: Patient, Example ARUP Accession: 25-139-401521 Patient Identifiers: 01234567890ABCD, 012345 Visit Number (FIN): 01234567890ABCD Page 1 of 2 | Printed: 6/9/2025 12:12:58 PM



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: Sequencing detects 76-98 percent of variants causing mild or moderate hemophilia A and 43-51 percent of variants causing severe hemophilia A. GENE TESTED: F8 (NM_000132.4) METHODOLOGY: Capture of all coding exons and exon-intron junctions of the F8 gene, followed by massively parallel sequencing. Sanger sequencing is performed as necessary to fill in regions of low coverage and confirm reported variants. ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced. LIMITATIONS: A negative result does not exclude a diagnosis of or carrier status for hemophilia A. This test only detects variants within the coding regions and intron-exon boundaries of the F8 gene. Variants in regions that are not included in the preferred transcript are not detected. This assay will not detect the common intron 22-A and intron 1 inversions. Regulatory region variants and deep intronic variants will not be identified. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. In males, lack of massively parallel sequencing coverage of one or more F8 exons may suggest the presence of a large deletion; however, this should be confirmed by a validated method. Diagnostic errors can occur due to rare sequence variations. In some cases, rvariants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpr

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	
Hemophilia A (F8) Specimen	25-139-401521	5/19/2025 12:06:00 PM	5/20/2025 7:54:05 AM	5/26/2025 10:00:00 PM	
Hemophilia A (F8) Interp	25-139-401521	5/19/2025 12:06:00 PM	5/20/2025 7:54:05 AM	5/26/2025 10:00:00 PM	

END OF CHART

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