

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 11/21/1985
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Hemophilia A (F8) 2 Inversions

ARUP test code 2001759

F8 Inv Specimen whole Blood

Hemophilia A (F8) Inversions Interp Negative

No pathogenic variants were detected in the F8 gene.

INTERPRETATION

Neither the factor 8 (F8) intron 22-A inversion nor the intron 1 inversion were detected. This negative result does not exclude carrier status for hemophilia A, as F8 variants undetected by this assay may be present. This result needs to be combined with F8 sequence analysis (ARUP test 2001747) for optimal pathogenic variant detection.

RECOMMENDATIONS

Medical management should rely on clinical findings. Genetic counseling is recommended.

COMMENTS

Reference Sequence: GenBank # NM_000132.3

This result has been reviewed and approved by Rong Mao, M.D.

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

BACKGROUND INFORMATION: Hemophilia A (F8) 2 Inversions

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 pathogenic variant.
PENETRANCE: 100 percent in males and 10 percent in females.
CAUSE: Pathogenic F8 gene variant.
CLINICAL SENSITIVITY: 51 percent of variants causing hemophilia A are detected by F8 inversion testing. This assay does not detect F8 variants 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 variants, other than the F8 intron 22-A and intron 1 inversions, will not be detected. Rare F8 intron 22-A and intron 1 inversions with different breakpoints may not be detected by this assay.

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 Inv Specimen	18-273-400059	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemophilia A (F8) Inversions Interp	18-273-400059	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