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PATIENT REPORT

Patient Age/Sex: 25 years Female

Specimen Collected: 29-Oct-25 14:18

Hemophilia A (F8) 2 Inversions |**Received: 29-Oct-25 14:19** **Report/Verified: 30-Oct-25 11:20**

Procedure	Result	Units	Reference Interval
F8 Inv Specimen	Whole Blood		
Hemophilia A (F8) Inversions Interp	**Intron 1** * f1 i1		

Result Footnote

f1: Hemophilia A (F8) Inversions Interp

INDICATION FOR TESTING
Carrier Status

RESULT
One pathogenic variant was detected in the F8 gene.

PATHOGENIC VARIANT
Gene: F8 (NM_000132.3)
Variant: Intron 1 inversion; Heterozygous

INTERPRETATION
One pathogenic inversion was detected in the factor 8 (F8) gene; thus this individual is a carrier of hemophilia A. 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. This individual's offspring have a 50 percent chance of inheriting the variant regardless of sex.

RECOMMENDATIONS
A baseline factor VIII clotting activity assay should be performed to determine if this individual is at increased risk for bleeding. Genetic consultation is indicated, including a discussion of medical screening and management. At-risk family members should be offered testing for the identified variant (Hemophilia A (F8) 2 Inversions, ARUP test code 2001759).

This result has been reviewed and approved by [redacted]

Test Information

i1: Hemophilia A (F8) Inversions Interp

BACKGROUND INFORMATION: Hemophilia A (F8) 2 Inversions

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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Test Information

i1: Hemophilia A (F8) Inversions Interp

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: 51 percent of variants causing severe 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/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. F8 variants, other than the F8 type 1 or type 2 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.

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.