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PATIENT REPORT

Patient Age/Sex: 24 years Female

Specimen Collected: 29-Oct-25 14:18

Hemophilia A (F8) 2 Inversions, Fetal | **Received: 29-Oct-25 14:18** **Report/Verified: 04-Nov-25 10:55**

Procedure	Result	Units	Reference Interval
Maternal Contamination Study Fetal Spec	Fetal Cells [f1]		
Maternal Contam Study,Maternal Spec	Whole Blood		
F8 INV FE Specimen	Cultured Amnio		
Hemophilia A (F8) Inversions Interp	**Intron 22** * [f2] [i1]		

Result Footnote

f1: Maternal Contamination Study Fetal Spec

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

f2: Hemophilia A (F8) Inversions Interp

INDICATION FOR TESTING
Prenatal Diagnosis

RESULT
One pathogenic variant was detected in the F8 gene.

PATHOGENIC VARIANT
Gene: F8 (NM_000132.3)
Variant: Intron 22-A inversion; Heterozygous

INTERPRETATION
According to information available to ARUP, the mother of this fetus was found to be a carrier of hemophilia A, harboring an F8 intron 22 inversion. The familial pathogenic intron 22 inversion was detected in the factor 8 (F8) gene in this prenatal sample; thus this female fetus is predicted to be 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
Genetic consultation is indicated. 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

Test Information

i1: Hemophilia A (F8) Inversions Interp

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: 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.