

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 7/11/1989
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Hemophilia A (F8) 2 Inversions, Fetal

ARUP test code 2001755

Maternal Contamination Study Fetal Spec

Fetal Cells

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

INTERPRETIVE INFORMATION: Maternal Cell Contamination, Fetal Specimen
Please refer to fetal report for interpretation.

Maternal Contam Study, Maternal Spec

whole Blood

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

F8 INV FE Specimen

Cultured Amnio

Hemophilia A (F8) Inversions Interp

Intron 22 *

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

RESULT

One pathogenic variant detected in the F8 gene.

DNA VARIANT

Pathogenic

Intron 22-A inversion; Hemizygous

INTERPRETATION

According to information provided to ARUP, the mother of this fetus carries the pathogenic intron 22-A inversion in the factor 8 (F8) gene. One copy of the pathogenic F8 intron 22-A gene inversion was identified in this prenatal sample; thus, this male fetus is predicted to be affected with hemophilia A.

RECOMMENDATIONS

Genetic consultation, including a discussion of medical screening and management, is indicated. Testing for the identified variant in other at-risk family members is recommended. For quality assurance purposes, ARUP Laboratories will confirm the above F8 inversion result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

COMMENTS

Reference Sequence: GenBank # NM_000132.3

Note: A familial positive control sample was tested.

This result has been reviewed and approved by Hunter Best, Ph.D.

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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	18-282-401954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	18-282-401954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
F8 INV FE Specimen	18-282-401954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemophilia A (F8) Inversions Interp	18-282-401954	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