

Hemophilia A F8 Gene, Intron 1 and 22 Inversion
Mutation Analysis, Prenatal

Patient ID SA00158465	Patient Name VALIDATIONSOF, TESTING F8INP	Birth Date 1985-02-05	Sex F	Age 38
Order Number SA00158465	Client Order Number SA00158465	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 28 Feb 2023 12:00		

HA F8 Int 1/22 Inversion, AF or CVS

F8INP Interpretation

1 MCR

These results do not provide evidence for a diagnosis of hemophilia A in the fetus of [MOTHER'S NAME] due to the intron 1/22 inversion mutation. This interpretation assumes that [MOTHER'S NAME] has previously been shown to carry the intron [1/22] inversion mutation in the F8 gene.

If [MOTHER'S NAME] has not been shown to be a carrier of the intron 1/22 inversion mutation, this result is not a true negative result. Testing of an affected or known carrier is recommended to determine the cause of hemophilia A in the family.

Errors in the information provided to us may lead to erroneous interpretation of test results.

A genetic consultation may be of benefit.

MCC Negative: Using a panel of polymorphic markers to compare maternal and fetal specimens, there was no evidence of maternal cell contamination in the fetal specimen.

ADDITIONAL INFORMATION

Genomic DNA is digested with Ksp22 I restriction enzyme, ligated with T4 DNA ligase, and amplified by PCR with primers

specific for the F8 intron 22 inversion mutations. Genomic DNA is amplified by PCR with primers specific for the F8 intron 1 inversion mutation.

HA F8 Int 1/22 Reason for Referral

MCR

Mother is a known or suspected carrier for Hemophilia A. Prenatal evaluation test for the presence of mutations in the F8 gene.

HA F8 Int 1/22 Inversion, AF or CVS

MCR

Negative

Reference Value
Not applicable

No apparent intron 1 or intron 22 inversions detected within the F8 gene.

HA F8 Intron 1/22 Reviewed By

MCR

JENNIFER MAIN

Received: 01 Mar 2023 10:12

Reported: 01 Mar 2023 10:30

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292