MAYO CLINIC LABORATORIES

1-800-533-1710

F8INV

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

Patient ID	Patient Name		Birth Date	Gender	Age
SA00065891	SAMPLEREPORT, F8INV		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00065891	SA00065891	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		10 Mar 2014 13:00			

HA F8 Intron 1/22 Inversion, B

F8INV Interpretation

MCR

This result is consistent with this individual being a carrier of hemophilia A. Hemophilia A is an X-linked condition; therefore, each male offspring of a carrier female has a 50% risk of being affected with hemophilia A.

Since we have documented the presence of a mutation within the F8 gene, carrier testing for other at risk individuals is possible.

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. Laboratory developed test.

HA F8 Int 1/22 Reason for Referral

MCR

Family history of Hemophilia A. Determine carrier status. Test for the presence of mutations in the F8 gene.

HA F8 Intron 1/22 Inversion, B

MCR

Intron 1 inversion detected

Reference Value
Not applicable

An intron 1 inversion was detected within the F8 gene. An intron 22 inversion was NOT detected.

HA F8 Intron 1/22 Reviewed By

MCR

Ann Strege

Received: 11 Mar 2014 14:08 **Reported:** 11 Mar 2014 15:09

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