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
SA00065890	SAMPLEREPORT, F8INV		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00065890	SA00065890	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		10 Mar 2014 13:00			

REVISED REPORT

HA F8 Intron 1/22 Inversion, B

F8INV Interpretation

MCR

The absence of an intron 1 or intron 22 inversion withthin the F8 gene does not rule out the possibility that this individual is a carrier of hemophilia A. Literature suggests that intron 22 inversions account for approximately 45%, and intron 1 inversions account for approximately 2–3%, of mutations associated with severe hemophilia (defined as having <1% factor VIII clotting acitvity).

Hemophilia A is an X-linked condition cause by mutations in the F8 gene (e.g. intron 1 and 22 inversions, point mutations, insertions, and deletions). Of note, this analysis evaluates for the presence of intron 1 and intron 22 inversions only. If testing is being performed because of a family history of hemophilia A, it is often helpful to test an obligate carrier or an affected family member. Indication of the mutation in this family would allow for more direct testing and risk assessment of at risk individuals.

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.

REVISED HA F8 Int 1/22 Reason for Referral

MCR

Patient has a diagnosis of Hemophilia A. Test for the presence of mutations in the F8 gene.

HA F8 Intron 1/22 Inversion, B

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

Ann Strege

Received: 11 Mar 2014 14:08 **Reported:** 12 Mar 2014 07:52

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

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REVISED REPORT

Previously Reported As:

HA F8 Intron 1/22 Inversion, B

HA F8 Int 1/22 Reason for Referral
Previously reported on 12 Mar 2014 at 07:51 as: MCR

see below

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