MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

FIXMS

Hemophilia B, F9 Gene Mutation Analysis, Whole Blood

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059002	SAMPLEREPORT, FIXMS-NORM		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059002	SA00059002	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		14 Jun 2013 16:01			

F9 Gene Mutation Screening, B

F9 Mutation Screen Interpretation

MCR

No mutations were found, suggesting that the patient is NOT a carrier for Hemophilia B.

This assay will not detect mutations known to cause congenital Hemophilia B outside the regions described in methods section and cannot exclude the possibility of somatic mosacism which has been reported in congenital Hemophilia B.

Large hemizygous deletions surrounding exons and large hemizygous deletions of F9 (procoagulant factor IX gene) will not be detected by this methodology in females.

It is often helpful to test an obligate carrier or an individual affected with Hemophilia B in this family. Identification of the mutation in this family would allow for more direct testing and risk assessment of at risk individuals.

Of patients with well established Hemophilia B, less than 1% will have no detectable deleterious mutations in F9 (procoagulant factor IX gene).

Genetic consultation may be of benefit for this individual and/or family to further discuss the implications of these findings.

ADDITIONAL INFORMATION

Direct mutation analysis of leukocyte genomic DNA performed by PCR amplification of the F9 gene, followed by fluorescent DNA sequencing analysis utilizing an Applied Biosystems Inc. (ABI) DNA Analyzer. Laboratory developed test.

F9 Mut Screen Reason for Referral

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Family history of Hemophilia B. Determine carrier status

F9 Mutation Screen Result

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Negative

Reference Value
Not applicable

No causative mutations were found within the regions described in the methods section of F9 (procoagulant factor IX gene).

F9 Mutation Screen Reviewed By

MCR

Tammy Bernatz

Received: 14 Jun 2013 16:04 **Reported:** 14 Jun 2013 16:11

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905