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
SA00059144	SAMPLEREPORT, FIXMS A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059144	SA00059144	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		17 Jun 2013 16:20			

F9 Gene Mutation Screening, B

F9 Mutation Screen Interpretation

MCR

The [describe the resulting mutation or amino acid change] mutation has been demonstrated in patients with [mild/moderate/severe] congenital Hemophilia B (procoagulant factor IX deficiency). Results therefore are consistent with a carrier state 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.

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

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

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 Mutation Screen Result

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Positive

Reference Value
Not applicable

A heterozygous [describe the nucleotide mutation], [describe the resulting mutation or amino acid change] was found in [describe the location] of F9 (procoagulant factor IX gene).

F9 Mut Screen Reason for Referral

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Patient has Hemophilia B. Analyze specimen for the presence of a detectable mutation.

F9 Mutation Screen Reviewed By

MCR

Tammy Bernatz

Received: 17 Jun 2013 16:21 **Reported:** 17 Jun 2013 16:26

Performing Site Legend

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