



Patient ID SA00057550	Patient Name SAMPLEREPORT, FIXKM	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00057550	Client Order Number SA00057550	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 16 May 2013 00:00		

F9 Gene Known Mutation, B

F9 Known Mutation Interpretation

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Hemophilia B due to [describe the nucleotide mutation] mutation resulting in [describe the resulting mutation or amino acid change] has previously been documented in this family.

The patient does NOT carry the [describe the resulting mutation or amino acid change] mutation.

Results therefore are consistent with the patient NOT being a carrier of Hemophilia B due to the [describe the resulting mutation or amino acid change] mutation.

Note: Since testing was focused on exon [exon number] of F9 (procoagulant factor IX gene), mutation(s) in other regions of functional significance in F9 cannot be excluded.

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 mosaicism 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.

Rarely, congenital Hemophilia B can be caused by double 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

Laboratory developed test.

F9 Known Mut Reason for Referral

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F9 Known Mutation Method

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Exon 1

Region analyzed includes exon 1 and splice junctions.

F9 Known Mutation Result

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Negative

Reference Value

Not applicable

No mutations were found within the coding region and splice junctions of exon [exon number] of F9 (procoagulant factor IX gene).

F9 Known Mutation Reviewed By

MCR

Tammy Bernatz

Received: 17 May 2013 14:35

Reported: 17 May 2013 14:37

Performing Site Legend

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