



Patient ID SA00057548	Patient Name SAMPLEREPOR, FIXKM	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00057548	Client Order Number SA00057548	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

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 in this patient.

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.

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) will not be detected by this methodology in females.

ADDITIONAL INFORMATION

Laboratory developed test.

F9 Known Mutation Result

MCR

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 Known Mut Reason for Referral

MCR

F9 Known Mutation Method

MCR

Exon 1

Region analyzed includes exon 1 and splice junctions.

F9 Known Mutation Reviewed By

MCR

Tammy Bernatz

Received: 17 May 2013 14:24

Reported: 17 May 2013 14:32

Performing Site Legend

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