MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

VWD2N

von Willebrand Disease 2N (Subtype Normandy), Blood

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059001	SAMPLEREPORT, VWD2N		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059001	SA00059001	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		13 Jun 2013 00:00			

von Willebrand Disease 2N (Normandy)

VWD2N Interpretation

MCR

This individual DOES have the Thr791Met mutation on ONE allele in Exon 18 of the vWF gene that encodes for the FVIII binding domain of vWF (Heterozygous mutant). Thr791Met mutation, when present in the heterozygous state, predicts a capacity of vWF to bind to FVIII that is intermediate between the wild type (normal) and homozygous state. Plasma FVIII activity is typically normal. The incidence of the individual known mutations causing von Willebrand disease (vWD) Type 2N are as follows: Thr791Met at 15%; Arg816Trp at 10%; and the Arg854Gln at >70%. These mutations account for >90% of the cases of vWD2N. This assay will not detect other mutations in the known FVIII binding domain of the vWF gene nor will it detect other mutations in the vWF gene. Consider genetic counseling and counseling of potentially affected family members regarding laboratory testing.

ADDITIONAL INFORMATION

This assay is performed by polymerase chain reaction (PCR) followed by restriction enzyme digestion to detect the three common mutations, Thr791Met (Tai1), Arg816Trp (AvaII) and Arg854Gln (Msp1). Laboratory developed test.

Thr791Met

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Heterozygous

Reference Value
Negative

Arg816Trp

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Negative

Reference Value
Negative

Arg854Gln

MCR

Negative

Reference Value
Negative

VWD2N Reviewed By

MCR

Tammy Bernatz

Received: 14 Jun 2013 16:03

Reported: 14 Jun 2013 16:07

Performing Site Legend

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