ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/8/2003
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

von Willebrand Disease, Type 2M (VWF) Sequencing

ARUP test code 2005490

vWD Type 2M (VWF) Sequencing Specimen: Whole Blood

vWD Type 2M (VWF) Sequencing Interp: Negative

TEST PERFORMED - 2005490
TEST DESCRIPTION - von Willebrand Disease Type 2M (VWF) Sequencing
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the VWF gene.

INTERPRETATION
No pathogenic variants were detected in the von Willebrand factor (VWF) gene using targeted sequencing of exons 28, 30, and 31. This decreases the risk for von Willebrand disease types 2A, 2B, and 2M by 80 percent.

RECOMMENDATIONS
Medical management should rely on clinical and phenotypic laboratory findings as well as family history. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000552.3 (VWF)
Nucleotide numbering begins at the "A" of the ATG initiation codon
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by [redacted]

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: von Willebrand Disease, Type 2M (VWF) Sequencing

CHARACTERISTICS: Mucocutaneous bleeding after brushing or flossing teeth, unexplained bruising, prolonged repeated nosebleeds, menorrhagia, and prolonged bleeding following childbirth, trauma or surgery.
INCIDENCE: Approximately 1 in 100 to 1 in 1000 individuals.
INHERITANCE: Autosomal dominant for type 2M.
PENETRANCE: Dominant mutations are incompletely penetrant when VWF:Ag and VWF:RCo levels are 25-50 IU/dL. Full penetrance is expected when VWF:Ag and VWF:RCo levels are less than 25 IU/dL.
CAUSE: Pathogenic VWF mutations in exons 28, 30, and 31.
CLINICAL SENSITIVITY: 80 percent for vWD types 2A, 2B, and 2M; unknown for other vWD subtypes.
METHODOLOGY: Bidirectional sequencing of VWF exons 28, 30, 31 and its intron-exon boundaries.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations, and large deletion/duplications will not be detected. Mutations lying outside of VWF exons 28, 30, and 31 will not be evaluated.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
vWD Type 2M (VWF) Sequencing Specimen	19-297-401142	10/23/2019 2:40:00 PM	10/24/2019 11:29:21 AM	11/13/2019 5:16:00 PM
vWD Type 2M (VWF) Sequencing Interp	19-297-401142	10/23/2019 2:40:00 PM	10/24/2019 11:29:21 AM	11/13/2019 5:16:00 PM

END OF CHART