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	3/5/1956
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

von Willebrand Disease, Type 2B (VWF) Sequencing

ARUP test code 2005486

vWD2B (VWF) Specimen	Whole Blood
vWD Type 2B (VWF) Sequencing Interp	Negative

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TEST PERFORMED - 2005486
TEST DESCRIPTION - von Willebrand Disease  Type 2B (VWF)
Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the VWF gene.

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

RECOMMENDATIONS
If this individual's phenotype is consistent with type 2B VWD
targeted testing of the GP1BA gene should be considered as the
phenotype of platelet-type VWD cannot be distinguished from type
2B VWD. 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]
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H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: von Willebrand Disease, Type 2B (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 types 2B, 2M and most of 2A; autosomal recessive for 20 percent of 2A.
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 exon 28.
CLINICAL SENSITIVITY: 80 percent for vWD types 2A, 2B, and 2M; unknown for other vWD subtypes.
METHODOLOGY: Bidirectional sequencing of VWF exon 28 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 exon 28 are not 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
vWD2B (VWF) Specimen	19-254-401577	9/9/2019 4:00 00 PM	9/11/2019 1:34:18 PM	9/20/2019 1:14:00 PM
vWD Type 2B (VWF) Sequencing Interp	19-254-401577	9/9/2019 4:00 00 PM	9/11/2019 1:34:18 PM	9/20/2019 1:14:00 PM

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