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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/30/2017
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

von Willebrand Disease, Platelet Type (GP1BA) 4 Mutations

ARUP test code 2005476

VWD Platelet Type (GP1BA) Seq Spcm	Whole Blood

VWD PlatletType (GP1BA) Seq Interp: Negative

TEST PERFORMED - 2005476
TEST DESCRIPTION - von Willebrand Disease Platelet Type (GP1BA) 4 Mutations
INDICATION FOR TEST - Confirm Diagnosis

RESULT
Negative for four pathogenic variants tested in the GP1BA gene.

INTERPRETATION
None of the 4 common GP1BA gene pathogenic variants targeted were detected; therefore, the risk for platelet type von Willebrand disease (PT-VWD) is reduced, but not eliminated. This individual may have a rare GP1BA pathogenic variant not identified by this assay.

RECOMMENDATIONS
This result should be correlated with VWF2A and VWF2B testing results (reported separately under ARUP accessions 19-177-402334 and 19-177-402287, respectively). If this individual has clinical findings consistent with PT-VWD, consider full gene sequencing of GP1BA. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000173.5 (GP1BA)
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, Platelet Type (GP1BA) 4 Mutations

CHARACTERISTICS: Mild to moderate mucocutaneous bleeding after brushing or flossing teeth, unexplained bruising, prolonged repeated nosebleeds, menorrhagia, and prolonged bleeding following childbirth, trauma or surgery. Thrombocytopenia may be present and worsen during the stress of severe infection, surgery or pregnancy.
INCIDENCE: Very rare.
INHERITANCE: Autosomal dominant.
CAUSE: Pathogenic GP1BA mutations.
CLINICAL SENSITIVITY: Unknown.
METHODOLOGY: Targeted bidirectional sequencing of the GP1BA gene mutations c.746G>T (p.Gly249Val), c.746G>A (p.Gly249Ser), and c.763A>G (p.Met255Val); PCR followed by fragment analysis of c.1306del27 (p.436_444del9).
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations, and large deletions/duplications will not be detected. GP1BA mutations other than the four targeted, 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 Platelet Type (GP1BA) Seq Spcm	19-177-402344	6/26/2019 2:12 00 PM	6/28/2019 3:22:53 AM	7/13/2019 4:57:00 PM
VWD PlatletType (GP1BA) Seq Interp	19-177-402344	6/26/2019 2:12 00 PM	6/28/2019 3:22:53 AM	7/13/2019 4:57:00 PM

END OF CHART

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