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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/1752
Gender:	Unknown
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	**Positive**	*

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

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

RESULT
One pathogenic variant was detected in the GP1BA gene.

DNA VARIANT
Classification: Pathogenic
Gene: GP1BA
Nucleic Acid Change: c.746G>T; Heterozygous
Amino Acid Alteration: p.Gly249Val

INTERPRETATION
One pathogenic GP1BA variant, c.746G>T; p.Gly249Val, was detected by targeted testing. This result is consistent with a diagnosis of platelet type von Willebrand disease (PT-VWD). This individual's offspring have a 50 percent risk of also being affected.

Evidence for variant classification: The GP1BA c.746G>T; p.Gly249Val variant (rs121908062), also known as Gly233Val, is described in the medical literature (Guerrero 2009, Miller 1991, Suva 2008), and classified as pathogenic in ClinVar (Variation ID: 4153). This variant is absent from the general population (Genome Aggregation Database), indicating it is not a common polymorphism. The glycine at codon 249 is highly conserved, and computational analyses predict this variant to be deleterious. Based on available information, this variant is considered to be pathogenic.

RECOMMENDATIONS
Genetic consultation, including a discussion of medical screening and management, is indicated. At-risk family members should be offered testing for the identified variant.

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.

REFERENCES
Guerrero JA et al. Visualizing the von Willebrand factor/glycoprotein Ib-IX axis with a platelet-type von Willebrand disease mutation. Blood. 2009 Dec 24;114(27):5541-6.
Miller JL et al. Mutation in the gene encoding the alpha chain of platelet glycoprotein Ib in platelet-type von Willebrand disease. Proc Natl Acad Sci U S A. 1991 Jun 1;88(11):4761-5.
Suva LJ et al. Platelet dysfunction and a high bone mass phenotype in a murine model of platelet-type von Willebrand disease. Am J Pathol. 2008 Feb;172(2):430-9.

This result has been reviewed and approved by [redacted]

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	19-336-112302	12/2/2019 1:09:00 PM	12/2/2019 1:14:19 PM	12/4/2019 8:52:00 AM
VWD PlatletType (GP1BA) Seq Interp	19-336-112302	12/2/2019 1:09:00 PM	12/2/2019 1:14:19 PM	12/4/2019 8:52:00 AM

END OF CHART