von Willebrand Disease, Type 2M (VWF) Sequencing
ARUP test code 2005490

<table>
<thead>
<tr>
<th>vWD Type 2M (VWF) Sequencing Specimen</th>
<th>Whole Blood</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>vWD Type 2M (VWF) Sequencing Interp</th>
<th>Negative</th>
</tr>
</thead>
</table>

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 Yuan Ji, Ph.D.
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.

INHERITANCE: Approximately 1 in 100 to 1 in 1000 individuals.

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

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Accession</th>
<th>Collected</th>
<th>Received</th>
<th>Verified/Reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>vWD Type 2M (VWF) Sequencing Specimen</td>
<td>19-297-401142</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
</tr>
<tr>
<td>vWD Type 2M (VWF) Sequencing Interp</td>
<td>19-297-401142</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
</tr>
</tbody>
</table>

END OF CHART