Quarterly HOTLINE: Effective January 4, 2019

New Test **3001129** **Capillary Malformation-Arteriovenous Malformation 2 (*EPHB4*) Sequencing** **EPHB4 FGS**

Available Now

Additional Technical Information

Patient History Form

Methodology: Polymerase Chain Reaction/Sequencing
Performed: Sun-Sat
Reported: Within 2 weeks

Specimen Required: Collect: Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD).
Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Refrigerated.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months

Interpretive Data:
Background Information for *Capillary Malformation-Arteriovenous Malformation 2* (*EPHB4*) Sequencing:
Characteristics of Capillary Malformation-Arteriovenous Malformation (CM-AVM): Multifocal, randomly distributed, capillary malformations (CM) that may be associated with a fast-flow lesion (arteriovenous malformations [AVM] or arteriovenous fistula). Fast-flow lesions in the skin, muscle, bone, or central nervous system can cause life-threatening complications such as bleeding, congestive heart failure, or neurological consequences. Capillary malformation-arteriovenous malformation syndrome type 1 (CM-AVM1) is caused by *RASA1* pathogenic variants; capillary malformation-arteriovenous malformation syndrome type 2 (CM-AVM2) is caused by *EPHB4* pathogenic variants.
Incidence: Estimated at 1 in 20,000 for CM-AVM1 and 1 in 12,000 for CM-AVM2.
Inheritance: Autosomal dominant.
Penetrance: 90-95 percent.
Cause: Pathogenic *EPHB4* or *RASA1* gene variants.
Gene Tested: *EPHB4* only.
Clinical Sensitivity: Not well established, at least 15 percent.
Methodology: Bidirectional sequencing of all coding regions and intron-exon boundaries of the *EPHB4* gene.
Analytical Specificity and Sensitivity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. Regulatory region variants, deep intronic variants, and large deletions/duplications will not be detected. Variants in genes other than *EPHB4* are not detected.

See Compliance Statement C: www.aruplab.com/CS

CPT Code(s): 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.