ARUP® LABORATORIES

Quarterly HOTLINE: Effective August 20, 2018

New Test	**3000531**	**Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy, CADASIL (*NOTCH3*), Sequencing**	**NOTCH3 FGS**

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

Specimen Required: <u>Collect:</u> Lavender (EDTA), Pink (K$_2$EDTA), or Yellow (ACD).
<u>Specimen Preparation:</u> Transport 3 mL whole blood. (Min: 1 mL)
<u>Storage/Transport Temperature:</u> Refrigerated.
<u>Stability (collection to initiation of testing):</u> Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months

Interpretive Data:
Background Information for Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy, CADASIL (*NOTCH3*), Sequencing:
Characteristics: Subcortical ischemic events, including transient ischemic attacks (TIAs) and strokes, are the most common presentation of CADASIL and present in approximately 85 percent of affected individuals. Cognitive defects and dementia are observed in 75 percent of affected individuals, migraines in 35 percent, psychiatric and mood disorders in 33 percent, and epilepsy in 10 percent. Age of onset and clinical presentation are highly variable.
Prevalence: 2-4 in 100,000.
Inheritance: Autosomal dominant.
Cause: Pathogenic variants in the *NOTCH3* gene.
Clinical Sensitivity: 95 percent.
Methodology: Bidirectional sequencing of *NOTCH3* coding regions and intron/exon boundaries.
Analytical Sensitivity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. Regulatory region variants and large deletion/duplications in the *NOTCH3* gene will not be detected.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

CPT Code(s): 81406

New York DOH approval pending. Call for status update.

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