Cerebral Cavernous Malformation: Deletion/Duplication Panel

Test Code: MD410
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Cerebral cavernous malformations are collections of small blood vessels in the brain that are enlarged and irregular in structure. This condition is inherited in an autosomal dominant manner, with pathogenic variants in three genes (CCM1, CCM2, and CCM3) accounting for 85-95% of all cases. While the exact function of these genes is not fully understood, studies show that the proteins produced from these genes are found in the junctions connecting neighboring blood vessel cells. Pathogenic variants in these genes impair the function of the protein complex, resulting in weakened cell-to-cell junctions and increased leakage from vessels as seen in cerebral cavernous malformations.

Reference:

Genes

CCM2, KRIT1, PDCD10

Indications

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of cerebral cavernous malformation.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood

Specimen Requirements:

- In EDTA (purple top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Type: Isolated DNA

Specimen Requirements:

- In microtainer: 10 ug

Isolation using the Qiagen Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight refrigeration.
Related Tests

- Cerebral Cavernous Malformation: Sequencing Panel