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.

**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:** DNA, Isolated

**Specimen Requirements:**  
Microtainer  
3µg  
Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**  
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

**Type:** Whole Blood (EDTA)

**Specimen Requirements:**  
EDTA (Purple Top)  
Infants and Young Children (2 years of age to 10 years old): 3-5 ml  
Older Children & Adults: 5-10 ml  
Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**  
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

### Related Tests

- Cerebral Cavernous Malformation: Sequencing Panel

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