Brugada Syndrome: Deletion/Duplication Panel

**Test Code:** MD095
**Turnaround time:** 2 weeks
**CPT Codes:** 81228 x1

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**Condition Description**

Brugada syndrome is characterized by cardiac conduction abnormalities. These cardiac abnormalities can result in sudden death. Often features such as syncope and/or arrhythmias present in adulthood; however, the age of diagnosis ranges from two days to 85 years. Pathogenic variants in eight genes are known to cause Brugada syndrome. Only 25% of individuals with Brugada syndrome have an identifiable pathogenic variant in one of the eight genes known to cause it. Most individuals with Brugada syndrome have an affected parent but approximately 1% of cases are the result of a de novo pathogenic variant.

Reference:
- GeneReviews

**Genes**

CACNA1C, CACNB2, GPD1L, HCN4, KCNE3, SCN1B, SCN3B, SCN5A

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of Brugada syndrome.
- Carrier testing in adults with a family history of Brugada syndrome.

**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: 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.

**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.

**Related Tests**

- Brugada Syndrome: Sequencing Panel
- Comprehensive Cardiovascular: Sequencing Panel
- Comprehensive Cardiovascular: Deletion/Duplication Panel