Brugada Syndrome: Deletion/Duplication Panel

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

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. 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:

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 delivery.
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