Arrhythmias: Deletion/Duplication Panel

Test Code: MD093
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Arrhythmia is any change from the normal sequence of electrical impulses of the heart. These impulses may happen too fast (tachycardia), too slowly (bradycardia), or erratically. Types of arrhythmias include atrial fibrillation, conduction disorders, premature contraction, ventricular fibrillation, tachycardia, and bradycardia. Arrhythmias can present with a broad spectrum of symptoms including palpitation, a fluttering sensation, fatigue, dizziness, lightheadedness, syncope, rapid heartbeat, shortness of breath, chest pain, and sudden cardiac arrest.

References:
- American Heart Association
- OMIM

Genes

AKAP9, ANK2, CACNA1C, CACNB2, CASQ2, DSC2, DSG2, DSP, GPD1L, HCN4, JUP, KCNE1, KCNE2, KCNE3, KCNH2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1, TMEM43

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of arrhythmia.
- Carrier testing in adults with a family history of arrhythmia.

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 pathogenic variants. 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:
Related Tests

- Comprehensive Cardiovascular: Sequencing Panel (MCAR1)
- Arrhythmias: Sequencing Panel