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
- KCNJ2
- KCNJ5
- KCNJ8
- KCNQ1
- NKX2-5
- PKP2
- RANGRF
- RYR2
- 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.

### 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: DNA, Isolated

**Specimen Requirements:**
- Microtainer
- 3µg

Isolation using the Perkin Elmer™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 24 hours of collection. Do not refrigerate or freeze.

### Related Tests

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