Sudden Cardiac Arrest (SCA): Deletion/Duplication Panel

Test Code: MD097
Turnaround time: 2 weeks
CPT Codes: 81414 x1

Condition Description

Sudden cardiac arrest is the abrupt loss of heart function due to a malfunction in the heart’s electrical system, such as an arrhythmia. The individual may or may not have been diagnosed with heart disease.

References:

- American Heart Association
- OMIM

Genes

ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, SCN5A

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of sudden cardiac arrest.
- Carrier testing in adults with a family history of sudden cardiac arrest.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that 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™ 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

- Comprehensive Cardiovascular Panel.
- Sudden Cardiac Arrest (SCA): Sequencing Panel.