Sudden Cardiac Arrest (SCA): Sequencing Panel

Test Code: MM097
Turnaround time: 6 weeks
CPT Codes: 81413 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, KCND3, 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

Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection

Next Generation Sequencing: Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Analytical Sensitivity: ~99%.

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: 60 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.

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
- Comprehensive Cardiovascular Panel.
- Sudden Cardiac Arrest (SCA): Deletion/Duplication Panel.