Arrhythmias: Sequencing Panel

Test Code: MM093
Turnaround time: 6 weeks
CPT Codes: 81404 x1, 81479 x1, 81403 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, CALM1, CASQ2, CTNNA3, DSC2, DSG2, DSP, GJA5, GPD1L, HCN4, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, NKX2-5, NPPA, PKP2, RANGRF, RYR2, SCN1B, SCN2B, SCN4B, SCN5A, SNTA1, TMEM43, TRDN, TRPM4

Indications

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

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

Clinical Sensitivity: Unknown. Pathogenic variants in the promoter region, some pathogenic variants in the introns and other regulatory element pathogenic variants 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 (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.

Type: DNA, Isolated
Specimen Requirements:
Microtainer
8µg
Isolation using the Perkin Elmer™Chemagen™ 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: Saliva

Specimen Requirements:
Oragene™ Saliva Collection Kit
Orangene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:
Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

Related Tests

- Comprehensive Cardiovascular: Sequencing Panel (MCAR1)
- Arrhythmias: Deletion/Duplication Panel