Long and Short QT Syndromes: Sequencing Panel

Test Code: MM094
Turnaround time: 6 weeks
CPT Codes: 81413 x1

Condition Description

Long QT syndrome (LQTS) is characterized by a QT interval that is prolonged on the surface electrocardiogram and a predisposition to early afterdepolarizations and torsades de pointes. LQTS can present clinically with palpitations, presyncope, syncope, or sudden cardiac death.

Short QT syndrome (SQTS) is characterized by an abnormally short QT interval and susceptibility to both ventricular tachyarrhythmias and atrial fibrillation.

References:
- OMIM

Genes

AKAP9, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN4B, SCN5A, SNTA1

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of Long QT syndrome (LQTS) and Short QT syndrome (SQTS).
- Carrier testing in adults with a family history of Long QT syndrome (LQTS) and Short QT syndrome (SQTS).

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. 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: 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
Oragene™ 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.

**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 cardiomyopathy panel
- Long and Short QT Syndromes: Deletion/Duplication Panel