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 after depolarizations 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, ANK2, 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: 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.
Related Tests

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