Long and Short QT Syndrome: Deletion/Duplication Panel

**Test Code:** MD094  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81414 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  
- 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

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

**Type: DNA, Isolated**

**Specimen Requirements:**
- Microtainer
- 3µg
- 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.

### Related Tests

- Comprehensive Cardiovascular: Sequencing Panel
- Comprehensive Cardiovascular: Deletion/Duplication Panel

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.
Long and Short QT Syndrome: Sequencing Panel