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. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

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

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: 10 ug
- Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.
- Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight.
Related Tests

- Comprehensive Cardiovascular: Sequencing Panel
- Comprehensive Cardiovascular: Deletion/Duplication Panel
- Long and Short QT Syndrome: Sequencing Panel