

Targeted Genes and Methodology Details for Long QT Syndrome Gene Panel

The following applies to LQTSG / Long QT Syndrome Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from November 2022 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
CACNA1C	NM_000719.7	-	-
CALM1	NM_006888.6	-	-
CALM2	NM_001743.6	-	-
CALM3	NM_005184.4	-	-
KCNE1	NM_000219.6	-	-
KCNH2	NM_000238.4	-	-
KCNJ2	NM_000891.3	-	-
KCNQ1	NM_000218.3	-	-
SCN5A	NM_198056.2	-	-
TRDN	NM_006073.4	chr6:123957870T>C (c.22+29A>G) chr6:123850462C>T (c.484+1189G>A)	CNV may not be detected in exons 7 and 22.