

# LONG QT SYNDROME PANEL<sup>1</sup> DG-4.1.0 (12 GENES)

<i>Gene</i>	<i>Twist X2 covered &gt;10x</i>	<i>Twist X2 covered &gt;20x</i>	<i>WGS covered &gt;10x</i>	<i>WGS covered &gt;20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
CACNA1C	100%	100%	100%	99.3%	Timothy syndrome, 601005; Long QT syndrome 8, 618447; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029; Brugada syndrome 3, 611875
CALM1	100%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916; Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100%	98.8%	Long QT syndrome 15, 616249
CALM3	100%	100%	100%	99.4%	Long QT syndrome 16, 618782; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
KCNE1	100%	100%	100%	99.8%	Jervell and Lange-Nielsen syndrome 2, 612347; Long QT syndrome 5, 613695

KCNE2	100%	100%	100%	99.5%	Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493
KCNH2	100%	100%	100%	98.1%	Short QT syndrome 1, 609620;Long QT syndrome 2, 613688
KCNJ2	100%	100%	100%	99.2%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNQ1	100%	99.7%	100%	98.1%	Short QT syndrome 2, 609621;Atrial fibrillation, familial, 3, 607554;Long QT syndrome 1, 192500;{Long QT syndrome 1, acquired, susceptibility to}, 192500;Jervell and Lange-Nielsen syndrome, 220400
SCN5A	100%	100%	100%	99.2%	Ventricular fibrillation, familial, 1, 603829;Heart block, progressive, type IA, 113900;Cardiomyopathy, dilated, 1E, 601154;Heart block, nonprogressive, 113900;Long QT syndrome 3, 603830;Sick sinus syndrome 1, 608567;Brugada syndrome 1, 601144;Atrial fibrillation, familial, 10, 614022;{Sudden infant death syndrome, susceptibility to}, 272120

TECRL	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TRDN	100%	100%	100%	99.7%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441

*Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*

*TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.*

*TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.*

*srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.*

*srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.*

*non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023.*

*This list is accurate for panel version DG 4.1.0*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*