

LONG QT SYNDROME PANEL¹ DG-4.3.0 (12 GENES)

<i>Gene</i>	<i>Twist X2 covered 10x</i>	<i>Twist X2 covered 20x</i>	<i>srWGS covered 10x</i>	<i>srWGS covered 15x</i>	<i>srWGS covered 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
CACNA1C	100%	100%	100%	99.9%	98.6%	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%	100%	99.5%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100%	99.8%	97.5%	Long QT syndrome 15, 616249
CALM3	100%	100%	100%	100%	99.3%	Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782

KCNE1	100%	100%	100%	100%	99.2%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNE2	100%	100%	100%	100%	99.4%	Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493
KCNH2	100%	100%	100%	99.7%	97.8%	Short QT syndrome 1, 609620;Long QT syndrome 2, 613688
KCNJ2	100%	100%	100%	99.8%	98.9%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNQ1	100%	99.6%	100%	99.5%	97.1%	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%	99.9%	98.5%	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%	99.9%	99.5%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TRDN	100%	100%	100%	100%	99.6%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441

Gene symbols used follow HGCN 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 mapped against GRCh38.

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 mapped against GRCh38.

srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.

srWGS 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 : November 25th, 2024.

This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors