

LONG QT SYNDROME PANEL¹ DG-5.0.0 (12 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
CACNA1C	100%	100%	100%	99.9%	99.4%	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%	99.9%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916; Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100%	99.9%	98.7%	Long QT syndrome 15, 616249
CALM3	100%	100%	100%	100%	99.9%	Long QT syndrome 16, 618782; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
KCNE1	100%	100%	100%	100%	99.7%	Jervell and Lange-Nielsen syndrome 2, 612347; Long QT syndrome 5, 613695
KCNE2	100%	100%	100%	100%	100%	Long QT syndrome 6, 613693; Atrial fibrillation, familial, 4, 611493
KCNH2	100%	100%	100%	99.9%	98.9%	Short QT syndrome 1, 609620; Long QT syndrome 2, 613688

KCNJ2	100%	100%	100%	100%	99.4%	Atrial fibrillation, familial, 9, 613980; Andersen syndrome, 170390; Short QT syndrome 3, 609622
KCNQ1	100%	100%	100%	99.9%	99%	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%	100%	99.5%	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	78.1%	78%	100%	100%	99.6%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TRDN	92.7%	92.7%	100%	100%	99.9%	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 5.0.0

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