

# WES LONG QT SYNDROME <sup>1</sup> DG 3.8.1

<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	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	99.6%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916; Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100.0%	98.9%	Long QT syndrome 15, 616249
CALM3	100.0%	100.0%	100.0%	99.4%	Long QT syndrome 16, 618782; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
KCNE1	100.0%	100.0%	100.0%	100.0%	Jervell and Lange-Nielsen syndrome 2, 612347; Long QT syndrome 5, 613695
KCNE2	100.0%	100.0%	100.0%	99.9%	Long QT syndrome 6, 613693; Atrial fibrillation, familial, 4, 611493
KCNH2	100.0%	100.0%	100.0%	98.8%	Short QT syndrome 1, 609620; Long QT syndrome 2, 613688
KCNJ2	100.0%	100.0%	100.0%	99.8%	Atrial fibrillation, familial, 9, 613980; Andersen syndrome, 170390; Short QT syndrome 3, 609622

KCNQ1	100.0%	100.0%	100.0%	99.6%	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	100.0%	100.0%	100.0%	99.4%	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	100.0%	100.0%	100.0%	99.4%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TRDN	99.9%	99.6%	100.0%	98.5%	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.

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 3.7.0.