

# LONG QT SYNDROME PANEL<sup>1</sup> DG-4.4.0 (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%              | 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%              | 100%              | 99.7%             | Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916; Long QT syndrome 14, 616247  |
| CALM2   | 73.5%                | 73.5%                | 100%              | 100%              | 99%               | Long QT syndrome 15, 616249   |
| CALM3   | 100%                 | 100%                 | 100%              | 100%              | 99.8%             | Long QT syndrome 16, 618782; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782  |
| KCNE1   | 100%                 | 100%                 | 100%              | 100%              | 99.8%             | Jervell and Lange-Nielsen syndrome 2, 612347; Long QT syndrome 5, 613695  |
| KCNE2   | 100%                 | 100%                 | 100%              | 100%              | 99.8%             | Long QT syndrome 6, 613693; Atrial fibrillation, familial, 4, 611493  |
| KCNH2   | 100%                 | 100%                 | 100%              | 99.9%             | 98.6%             | Short QT syndrome 1, 609620; Long QT syndrome 2, 613688   |

|       |      |      |      |       |       |   |
|-------|------|------|------|-------|-------|---|
| KCNJ2 | 100% | 100% | 100% | 100%  | 99.7% | Atrial fibrillation, familial, 9, 613980; Andersen syndrome, 170390; Short QT syndrome 3, 609622  |
| KCNQ1 | 100% | 100% | 100% | 100%  | 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% | 99.9% | 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% | 100%  | 99.9% | Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021   |
| TRDN  | 100% | 100% | 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 4.4.0*

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