

LONG QT SYNDROME

GENE PANEL DG 3.6.0 (10 GENES)

Releasedate: 05-04-2023

<i>Gene</i>	<i>TWIST X2 covered >10x</i>	<i>TWIST X2 covered >20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
CACNA1C	100%	100%	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%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 Long QT syndrome 14, 616247
CALM2	73%	73%	Long QT syndrome 15, 616249
CALM3	100%	100%	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
KCNE1	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	100%	100%	Long QT syndrome 6, 613693 Atrial fibrillation, familial, 4, 611493
KCNH2	100%	100%	Short QT syndrome 1, 609620 Long QT syndrome 2, 613688
KCNQ1	100%	100%	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
SCN5A	100%	100%	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
TRDN	99%	99%	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.

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TWIST X2 is the chemistry used for WES analysis.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with coverage denoting NC are non-protein coding genes.

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 3.6.0

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