ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL[1] DG-5.0.0 (31 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
CASQ2	87.6%	87.5%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
DES	92.6%	92.6%	100%	99.9%	99.1%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419

DPP6	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	100%	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	88%	88%	100%	100%	99.6%	Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193
DSP	99.4%	99.3%	100%	100%	99.5%	Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
FLNC	100%	100%	100%	100%	99.7%	Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524

GNB2	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
HCN4	100%	100%	100%	100%	98.8%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
JUP	100%	100%	100%	99.9%	99.1%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
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

LMNA	100%	100%	100%	100%	99.5%	Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112
PKP2	99.7%	98.4%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLN	100%	100%	100%	100%	100%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874
PPA2	82.8%	82.6%	100%	100%	99.9%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
RYR2	99.8%	99.8%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000

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
SLC4A3	100%	100%	100%	100%	99.2%	Short QT syndrome 7, 620231
TECRL	78.1%	78%	100%	100%	99.6%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TMEM43	100%	100%	100%	100%	99.4%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TNNI3K	100%	100%	100%	100%	99.8%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	100%	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494

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.
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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