ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS GENE PANEL DG 3.5.0 (26 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
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	74%	74%	Long QT syndrome 15, 616249
CALM3	100%	100%	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CASQ2	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
DES	100%	100%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DSC2	100%	100%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	100%	100%	Cardiomyopathy, dilated, 1BB, 612877 Arrhythmogenic right ventricular dysplasia 10, 610193
DSP	100%	100%	Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 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%	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524

GNB2	100%	100%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 ?Sick sinus syndrome 4, 619464
HCN4	100%	100%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
JUP	100%	100%	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528
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
KCNJ2	100%	100%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KCNQ1	100%	100%	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
LMNA	100%	100%	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	100%	99%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLN	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
RYR2	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 Arrhythmogenic right ventricular dysplasia 2, 600996
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
TMEM43	100%	100%	Arrhythmogenic right ventricular dysplasia 5, 604400 Auditory neuropathy, autosomal dominant 3, 619832 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TNNT2	100%	100%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, hypertrophic, 2, 115195 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TRDN	100%	100%	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.
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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 : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors