

ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL¹ 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, 11, 604765; Myopathy, myofibrillar, 1, 601419 |

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| 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 |

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| 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 |

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| 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 |

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| 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 |

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| TRDN | 92.7% | 92.7% | 100% | 100% | 99.9% | Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441 |
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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 5.0.0

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