

HEART DISORDERS PANEL¹ DG-3.9.0 (334 GENES)

| <i>Gene</i> | <i>Twist X2 covered >10x</i> | <i>Twist X2 covered >20x</i> | <i>WGS covered >10x</i> | <i>WGS covered >20x</i> | <i>Associated Phenotype description and OMIM disease ID</i> |
|-------------|---------------------------------|---------------------------------|----------------------------|----------------------------|--|
| AARS2 | 100.0% | 100.0% | 100.0% | 99.4% | Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096 |
| ABCC6 | 100.0% | 100.0% | 100.0% | 99.3% | Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850 |
| ABCC9 | 100.0% | 100.0% | 100.0% | 98.4% | Cardiomyopathy, dilated, 10, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719 |

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| ABL1 | 100.0% | 100.0% | 100.0% | 99.4% | Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232; Congenital heart defects and skeletal malformations syndrome, 617602 |
| ACAD8 | 100.0% | 100.0% | 100.0% | 99.1% | Isobutyryl-CoA dehydrogenase deficiency, 611283 |
| ACAD9 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| ACADVL | 100.0% | 100.0% | 99.9% | 96.4% | VLCAD deficiency, 201475 |
| ACSF3 | 100.0% | 100.0% | 100.0% | 98.8% | Combined malonic and methylmalonic aciduria, 614265 |
| ACTA1 | 100.0% | 100.0% | 100.0% | 97.1% | Congenital myopathy 2B, severe infantile, autosomal recessive, 620265; ?Myopathy, scapulohumeroperoneal, 616852; Congenital myopathy 2C, severe infantile, autosomal dominant, 620278; Congenital myopathy 2A, typical, autosomal dominant, 161800 |

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| ACTC1 | 100.0% | 100.0% | 100.0% | 99.5% | Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424 |
| ACTN2 | 100.0% | 100.0% | 99.9% | 97.9% | Myopathy, distal, 6, adult onset, 618655;Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158;Congenital myopathy 8, 618654;Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 |
| ACVR2B | 100.0% | 100.0% | 100.0% | 98.7% | Heterotaxy, visceral, 4, autosomal, 613751 |
| ADAMTS19 | 100.0% | 100.0% | 100.0% | 98.9% | Cardiac valvular dysplasia 2, 620067 |
| ADCY5 | 100.0% | 99.9% | 100.0% | 97.4% | Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647 |
| ADNP | 100.0% | 100.0% | 100.0% | 98.7% | Helsmoortel-van der Aa syndrome, 615873 |

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| AGK | 91.7% | 91.7% | 100.0% | 98.9% | Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350 |
| AGL | 100.0% | 100.0% | 100.0% | 98.1% | Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400 |
| AGPAT2 | 100.0% | 100.0% | 100.0% | 97.9% | Lipodystrophy, congenital generalized, type 1, 608594 |
| AKAP9 | 100.0% | 100.0% | 100.0% | 97.4% | ?Long QT syndrome 11, 611820 |
| ALDH1A2 | 100.0% | 99.9% | 100.0% | 98.4% | Diaphragmatic hernia 4, with cardiovascular defects, 620025 |
| ALMS1 | 100.0% | 100.0% | 100.0% | 98.4% | Alstrom syndrome, 203800 |
| ALPK3 | 100.0% | 100.0% | 100.0% | 98.3% | Cardiomyopathy, familial hypertrophic 27, 618052 |
| ANK2 | 100.0% | 100.0% | 100.0% | 98.6% | Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919 |
| ANKRD1 | 100.0% | 99.9% | 100.0% | 96.2% | |
| ANKRD11 | 100.0% | 100.0% | 100.0% | 98.0% | KBG syndrome, 148050 |
| ATPAF2 | 100.0% | 100.0% | 100.0% | 99.1% | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 |
| BAG3 | 100.0% | 100.0% | 100.0% | 98.2% | Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954 |
| BANF1 | 100.0% | 100.0% | 100.0% | 97.3% | Nestor-Guillermo progeria syndrome, 614008 |

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| BICD2 | 100.0% | 100.0% | 100.0% | 99.1% | Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 |
| BMPR2 | 100.0% | 99.8% | 100.0% | 99.0% | Pulmonary hypertension, familial primary, 1, with or without HHT, 178600;Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600;Pulmonary venoocclusive disease 1, 265450 |
| BRAF | 100.0% | 100.0% | 99.9% | 96.7% | Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Non-small cell lung cancer, somatic, 211980 |

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| BSCL2 | 100.0% | 100.0% | 100.0% | 99.3% | Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924 |
| BVES | 100.0% | 100.0% | 100.0% | 98.4% | Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 |
| CACNA1C | 100.0% | 100.0% | 100.0% | 99.0% | 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 |
| CACNA1D | 100.0% | 100.0% | 100.0% | 98.5% | Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896 |
| CACNA2D1 | 100.0% | 100.0% | 100.0% | 97.1% | Developmental and epileptic encephalopathy 110, 620149 |

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| CACNB2 | 100.0% | 100.0% | 100.0% | 97.0% | Brugada syndrome 4, 611876 |
| CALM1 | 100.0% | 100.0% | 100.0% | 99.6% | Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247 |
| CALM2 | 73.5% | 73.5% | 100.0% | 97.3% | Long QT syndrome 15, 616249 |
| CALM3 | 100.0% | 100.0% | 100.0% | 98.8% | Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782 |
| CASQ2 | 100.0% | 100.0% | 100.0% | 98.6% | Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 |
| CASZ1 | 99.7% | 98.9% | 99.9% | 96.7% | |
| CAV1 | 100.0% | 100.0% | 100.0% | 99.2% | Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721 |

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| CAV3 | 100.0% | 100.0% | 100.0% | 99.7% | Myopathy, distal, Tateyama type, 614321;Creatine phosphokinase, elevated serum, 123320;Cardiomyopathy, familial hypertrophic, 192600;Rippling muscle disease 2, 606072;Long QT syndrome 9, 611818 |
| CCDC114 | 100.0% | 100.0% | 100.0% | 98.9% | Ciliary dyskinesia, primary, 20, 615067 |
| CDH2 | 100.0% | 100.0% | 100.0% | 99.0% | Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 |
| CFAP45 | 100.0% | 100.0% | 100.0% | 98.7% | Heterotaxy, visceral, 11, autosomal, with male infertility, 619608 |
| CFAP53 | 100.0% | 100.0% | 99.9% | 97.2% | Heterotaxy, visceral, 6, autosomal recessive, 614779 |
| CFC1 | 100.0% | 100.0% | 100.0% | 99.6% | Heterotaxy, visceral, 2, autosomal, 605376 |
| CHD4 | 100.0% | 100.0% | 100.0% | 98.2% | Sifrim-Hitz-Weiss syndrome, 617159 |

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| CHD7 | 100.0% | 100.0% | 100.0% | 98.6% | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800 |
| CHKB | 100.0% | 100.0% | 100.0% | 98.7% | Muscular dystrophy, congenital, megaconial type, 602541 |
| CHRM2 | 99.1% | 98.1% | 100.0% | 99.1% | |
| CITED2 | 100.0% | 100.0% | 100.0% | 96.5% | Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431 |
| COL3A1 | 100.0% | 100.0% | 100.0% | 98.1% | Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343 |
| COQ2 | 96.3% | 96.3% | 100.0% | 98.5% | {Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426 |
| COX15 | 100.0% | 100.0% | 100.0% | 98.5% | Mitochondrial complex IV deficiency, nuclear type 6, 615119 |
| CPT1A | 100.0% | 100.0% | 100.0% | 98.5% | CPT deficiency, hepatic, type IA, 255120 |

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|--------|--------|--------|--------|-------|---|
| CPT2 | 100.0% | 100.0% | 100.0% | 98.7% | {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110 |
| CRELD1 | 100.0% | 100.0% | 100.0% | 98.7% | Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217 |
| CRYAB | 100.0% | 100.0% | 100.0% | 99.1% | Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869;Myopathy, myofibrillar, 2, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184 |
| CSRP3 | 100.0% | 100.0% | 100.0% | 99.7% | ?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124 |
| CTNNA3 | 99.9% | 99.8% | 100.0% | 98.4% | Arrhythmogenic right ventricular dysplasia 13, 615616 |

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| CTNND1 | 100.0% | 100.0% | 100.0% | 98.7% | Blepharocheilodontic syndrome 2, 617681 |
| DCHS1 | 100.0% | 100.0% | 100.0% | 99.7% | Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390 |
| DES | 100.0% | 100.0% | 100.0% | 98.9% | Scapulooperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419 |
| DMD | 99.5% | 99.1% | 97.7% | 70.9% | Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200 |
| DNAJC19 | 100.0% | 100.0% | 100.0% | 98.2% | 3-methylglutaconic aciduria, type V, 610198 |
| DOLK | 100.0% | 100.0% | 100.0% | 98.1% | Congenital disorder of glycosylation, type 1m, 610768 |
| DOT1L | 100.0% | 100.0% | 100.0% | 99.4% | |
| DPM3 | 100.0% | 100.0% | 100.0% | 94.8% | ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 |

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| DPP6 | 100.0% | 99.9% | 100.0% | 98.2% | Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956 |
| DSC2 | 100.0% | 100.0% | 100.0% | 98.6% | Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476 |
| DSG2 | 100.0% | 100.0% | 100.0% | 99.0% | Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193 |
| DSP | 100.0% | 100.0% | 100.0% | 98.0% | 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 |

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|--------|--------|--------|--------|-------|--|
| DTNA | 100.0% | 100.0% | 100.0% | 98.8% | Left ventricular noncompaction 1, with or without congenital heart defects, 604169 |
| DYRK1A | 100.0% | 100.0% | 100.0% | 98.7% | Intellectual developmental disorder, autosomal dominant 7, 614104 |
| DZIP1 | 100.0% | 100.0% | 100.0% | 97.1% | Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840 |
| EEF1A2 | 100.0% | 100.0% | 100.0% | 98.7% | Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393 |
| EHMT1 | 100.0% | 99.9% | 99.9% | 98.9% | Kleefstra syndrome 1, 610253 |
| ELAC2 | 100.0% | 100.0% | 100.0% | 99.3% | {Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440 |
| ELN | 100.0% | 100.0% | 100.0% | 98.8% | Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500 |
| EMD | 100.0% | 99.5% | 98.1% | 71.4% | Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 |

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|--------|--------|--------|--------|-------|---|
| ENPP1 | 100.0% | 99.7% | 100.0% | 97.7% | {Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522 |
| FAH | 100.0% | 100.0% | 100.0% | 98.5% | Tyrosinemia, type I, 276700 |
| FBN1 | 100.0% | 100.0% | 100.0% | 99.1% | Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900 |
| FBN2 | 100.0% | 100.0% | 100.0% | 99.4% | Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050 |
| FBXO32 | 100.0% | 100.0% | 100.0% | 99.5% | |
| FGF12 | 100.0% | 100.0% | 100.0% | 99.7% | Developmental and epileptic encephalopathy 47, 617166 |

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| FHL1 | 100.0% | 99.9% | 97.9% | 69.1% | Myopathy, X-linked, with postural muscle atrophy, 300696;Emery-Dreifuss muscular dystrophy 6, X-linked, 300696;?Uruguay faciocardiomusculoskeletal syndrome, 300280;Scapulooperoneal myopathy, X-linked dominant, 300695;Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718;Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 |
| FHL2 | 100.0% | 100.0% | 100.0% | 99.5% | |
| FHOD3 | 100.0% | 100.0% | 100.0% | 98.3% | Cardiomyopathy, familial hypertrophic, 28, 619402 |

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| FKRP | 100.0% | 100.0% | 100.0% | 99.9% | Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 |
| FKTN | 100.0% | 100.0% | 99.9% | 98.0% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 |
| FLII | 100.0% | 100.0% | 100.0% | 99.1% | Cardiomyopathy, dilated, 2J, 620635 |

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|------|--------|--------|--------|-------|---|
| FLNA | 100.0% | 99.9% | 99.0% | 78.6% | Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620 |
| FLNC | 100.0% | 100.0% | 100.0% | 99.4% | Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524 |
| FLT4 | 100.0% | 100.0% | 100.0% | 99.2% | Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780 |

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| FNIP1 | 100.0% | 100.0% | 100.0% | 98.8% | Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 |
| FOXC2 | 100.0% | 100.0% | 99.9% | 92.9% | Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 |
| FOXD4 | 100.0% | 100.0% | 100.0% | 99.1% | |
| FOXH1 | 100.0% | 100.0% | 100.0% | 99.2% | |
| FOXJ1 | 100.0% | 100.0% | 100.0% | 97.2% | Ciliary dyskinesia, primary, 43, 618699 |
| FOXL1 | 100.0% | 100.0% | 99.9% | 95.1% | Otosclerosis 11, 620576 |
| GAA | 100.0% | 100.0% | 100.0% | 99.5% | Glycogen storage disease II, 232300 |
| GATA4 | 100.0% | 100.0% | 99.8% | 96.0% | Tetralogy of Fallot, 187500;Atrial septal defect 2, 607941;Ventricular septal defect 1, 614429;Atrioventricular septal defect 4, 614430;?Testicular anomalies with or without congenital heart disease, 615542 |
| GATA5 | 100.0% | 100.0% | 100.0% | 97.6% | Congenital heart defects, multiple types, 5, 617912 |

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| GATA6 | 100.0% | 100.0% | 100.0% | 92.6% | Atrial septal defect 9, 614475;Persistent truncus arteriosus, 217095;Pancreatic agenesis and congenital heart defects, 600001;Atrioventricular septal defect 5, 614474;Tetralogy of Fallot, 187500 |
| GATAD1 | 100.0% | 100.0% | 100.0% | 98.3% | ?Cardiomyopathy, dilated, 2B, 614672 |
| GATB | 100.0% | 100.0% | 100.0% | 99.1% | ?Combined oxidative phosphorylation deficiency 41, 618838 |
| GATC | 100.0% | 100.0% | 100.0% | 99.1% | Combined oxidative phosphorylation deficiency 42, 618839 |
| GBE1 | 100.0% | 99.9% | 100.0% | 98.4% | Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570 |
| GDF1 | 100.0% | 100.0% | 100.0% | 99.5% | Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530 |
| GDF2 | 100.0% | 100.0% | 100.0% | 99.5% | Telangiectasia, hereditary hemorrhagic, type 5, 615506 |

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| GJA5 | 100.0% | 100.0% | 100.0% | 99.8% | Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770 |
| GLA | 90.9% | 90.9% | 98.3% | 73.7% | Fabry disease, cardiac variant, 301500;Fabry disease, 301500 |
| GLB1 | 100.0% | 100.0% | 100.0% | 98.9% | GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600 |
| GLIS1 | 100.0% | 100.0% | 100.0% | 99.5% | |
| GLYR1 | 100.0% | 100.0% | 100.0% | 98.6% | |
| GMPPB | 100.0% | 100.0% | 100.0% | 99.4% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 |

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| GNB2 | 100.0% | 100.0% | 100.0% | 99.8% | Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464 |
| GNPTAB | 100.0% | 100.0% | 100.0% | 98.5% | Mucopolipidosis III alpha/beta, 252600;Mucopolipidosis II alpha/beta, 252500 |
| GPD1L | 100.0% | 100.0% | 100.0% | 97.7% | Brugada syndrome 2, 611777 |
| HADHA | 100.0% | 100.0% | 100.0% | 98.8% | HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016 |
| HADHB | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial trifunctional protein deficiency 2, 620300 |
| HAND1 | 100.0% | 100.0% | 100.0% | 98.2% | |
| HAND2 | 100.0% | 100.0% | 98.3% | 73.0% | |
| HCN2 | 94.4% | 92.1% | 93.1% | 78.6% | Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477 |
| HCN3 | 100.0% | 100.0% | 100.0% | 99.0% | |

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| HCN4 | 100.0% | 100.0% | 100.0% | 96.9% | Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123 |
| HEY2 | 100.0% | 100.0% | 100.0% | 98.1% | |
| HFE | 100.0% | 100.0% | 100.0% | 98.3% | Hemochromatosis, type 1, 235200 |
| HJV | 100.0% | 100.0% | 100.0% | 98.6% | Hemochromatosis, type 2A, 602390 |
| HSPB6 | 100.0% | 100.0% | 99.9% | 95.3% | |
| HSPD1 | 100.0% | 100.0% | 100.0% | 98.8% | Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233 |
| IDUA | 100.0% | 100.0% | 100.0% | 97.9% | Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014 |
| ILK | 100.0% | 100.0% | 100.0% | 99.3% | |
| ITGA7 | 100.0% | 100.0% | 100.0% | 99.0% | Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 |
| ITPA | 100.0% | 100.0% | 100.0% | 97.8% | [Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647 |

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|---------|--------|--------|--------|-------|--|
| JAG1 | 100.0% | 100.0% | 100.0% | 99.6% | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500 |
| JPH2 | 100.0% | 99.9% | 100.0% | 99.1% | Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873 |
| JUP | 100.0% | 100.0% | 100.0% | 99.4% | Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528 |
| KBTBD13 | 100.0% | 100.0% | 100.0% | 99.4% | Nemaline myopathy 6, autosomal dominant, 609273 |
| KCNA5 | 100.0% | 100.0% | 100.0% | 98.7% | Atrial fibrillation, familial, 7, 612240 |
| KCND2 | 99.9% | 99.3% | 100.0% | 98.2% | |
| KCND3 | 100.0% | 100.0% | 100.0% | 99.3% | Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399 |
| KCNE1 | 100.0% | 100.0% | 100.0% | 99.7% | Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695 |
| KCNE2 | 100.0% | 100.0% | 100.0% | 99.8% | Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493 |

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|--------|--------|--------|--------|-------|---|
| KCNE3 | 100.0% | 100.0% | 100.0% | 99.8% | ?Brugada syndrome 6, 613119 |
| KCNE4 | 100.0% | 100.0% | 100.0% | 99.3% | |
| KCNE5 | 100.0% | 99.9% | 98.5% | 72.5% | |
| KCNH2 | 100.0% | 100.0% | 100.0% | 96.9% | Short QT syndrome 1, 609620;Long QT syndrome 2, 613688 |
| KCNJ11 | 100.0% | 100.0% | 100.0% | 99.7% | Diabetes, permanent neonatal 2, with or without neurologic features, 618856;{Diabetes mellitus, type 2, susceptibility to}, 125853;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820 |
| KCNJ2 | 100.0% | 100.0% | 100.0% | 99.4% | Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622 |
| KCNJ5 | 100.0% | 100.0% | 100.0% | 99.0% | Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677 |
| KCNJ8 | 100.0% | 100.0% | 100.0% | 99.5% | |
| KCNK3 | 100.0% | 100.0% | 100.0% | 96.8% | Pulmonary hypertension, primary, 4, 615344 |
| KCNN3 | 100.0% | 100.0% | 100.0% | 98.1% | Zimmermann-Laband syndrome 3, 618658 |

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|--------|--------|--------|--------|-------|--|
| KCNQ1 | 100.0% | 100.0% | 100.0% | 97.2% | 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 |
| KDR | 100.0% | 100.0% | 100.0% | 98.5% | {Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089 |
| KLHL24 | 100.0% | 100.0% | 100.0% | 99.5% | Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294 |
| KMT2A | 100.0% | 100.0% | 100.0% | 97.9% | Wiedemann-Steiner syndrome, 605130 |
| KMT2D | 100.0% | 100.0% | 100.0% | 98.8% | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920 |

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|-------|--------|--------|--------|-------|--|
| KRAS | 100.0% | 100.0% | 100.0% | 99.7% | Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800 |
| LAMA2 | 99.8% | 99.5% | 100.0% | 99.0% | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 |
| LAMA4 | 100.0% | 100.0% | 100.0% | 99.1% | Cardiomyopathy, dilated, 1JJ, 615235 |
| LAMP2 | 100.0% | 100.0% | 98.0% | 72.4% | Danon disease, 300257 |

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|--------|--------|--------|--------|-------|---|
| LDB3 | 100.0% | 100.0% | 100.0% | 98.6% | Left ventricular noncompaction 3, 601493;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 |
| LEFTY2 | 100.0% | 100.0% | 100.0% | 99.5% | |
| LEMD2 | 100.0% | 100.0% | 100.0% | 95.9% | Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500 |
| LIMS2 | 100.0% | 100.0% | 100.0% | 99.3% | ?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 |
| LMCD1 | 100.0% | 100.0% | 99.9% | 98.4% | |

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|--------|--------|--------|--------|-------|---|
| LMNA | 100.0% | 100.0% | 100.0% | 99.1% | 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 |
| LMOD2 | 100.0% | 100.0% | 99.9% | 95.1% | Cardiomyopathy, dilated, 2G, 619897 |
| LRRC10 | 100.0% | 100.0% | 100.0% | 99.6% | |
| LZTR1 | 100.0% | 100.0% | 100.0% | 99.7% | Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670 |

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|--------|--------|--------|--------|-------|--|
| MED13L | 100.0% | 99.6% | 100.0% | 98.5% | Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 |
| MIB1 | 100.0% | 100.0% | 100.0% | 99.2% | Left ventricular noncompaction 7, 615092 |
| MIPEP | 100.0% | 100.0% | 100.0% | 98.7% | Combined oxidative phosphorylation deficiency 31, 617228 |
| MLYCD | 100.0% | 100.0% | 100.0% | 97.4% | Malonyl-CoA decarboxylase deficiency, 248360 |
| MMP21 | 100.0% | 100.0% | 100.0% | 98.4% | Heterotaxy, visceral, 7, autosomal, 616749 |
| MTO1 | 93.7% | 91.1% | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 10, 614702 |
| MUC16 | 100.0% | 100.0% | 100.0% | 99.3% | |
| MYBPC3 | 100.0% | 100.0% | 100.0% | 99.7% | Cardiomyopathy, hypertrophic, 4, 115197;Cardiomyopathy, dilated, 1MM, 615396;Left ventricular noncompaction 10, 615396 |
| MYBPHL | 100.0% | 100.0% | 100.0% | 99.6% | |
| MYH11 | 100.0% | 100.0% | 100.0% | 98.1% | Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350 |

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| MYH6 | 100.0% | 100.0% | 100.0% | 97.7% | {Sick sinus syndrome 3}, 614090;Atrial septal defect 3, 614089;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251 |
| MYH7 | 100.0% | 100.0% | 100.0% | 99.0% | Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358 |
| MYH7B | 100.0% | 100.0% | 100.0% | 99.1% | |
| MYL2 | 100.0% | 100.0% | 100.0% | 98.9% | Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile- onset, with cardiomyopathy, 619424 |
| MYL3 | 100.0% | 100.0% | 100.0% | 99.3% | Cardiomyopathy, hypertrophic, 8, 608751 |
| MYL4 | 100.0% | 100.0% | 100.0% | 99.6% | ?Atrial fibrillation, familial, 18, 617280 |
| MYL7 | 100.0% | 100.0% | 100.0% | 98.2% | |

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|-------|--------|--------|--------|-------|--|
| MYLK3 | 100.0% | 100.0% | 100.0% | 99.1% | |
| MYO6 | 100.0% | 100.0% | 100.0% | 97.9% | Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821 |
| MYOM1 | 100.0% | 100.0% | 100.0% | 98.5% | |
| MYOT | 100.0% | 100.0% | 100.0% | 98.3% | Myopathy, myofibrillar, 3, 609200 |
| MYOZ2 | 100.0% | 100.0% | 100.0% | 99.0% | Cardiomyopathy, hypertrophic, 16, 613838 |
| MYPN | 100.0% | 100.0% | 100.0% | 98.9% | Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248 |
| MYRF | 100.0% | 100.0% | 100.0% | 98.6% | Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280 |

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| NAA15 | 96.6% | 96.6% | 100.0% | 98.3% | Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 |
| NDUFB11 | 99.7% | 97.9% | 88.0% | 60.9% | Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021 |
| NEBL | 99.8% | 99.2% | 100.0% | 97.9% | |
| NEXN | 100.0% | 100.0% | 99.9% | 94.7% | Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876 |
| NF1 | 100.0% | 100.0% | 100.0% | 98.6% | Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321 |

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|--------|--------|--------|--------|-------|---|
| NKX2-5 | 100.0% | 100.0% | 100.0% | 98.2% | Hypoplastic left heart syndrome 2, 614435;Tetralogy of Fallot, 187500;Hypothyroidism, congenital nongoitrous, 5, 225250;Conotruncal heart malformations, variable, 217095;Ventricular septal defect 3, 614432;Atrial septal defect 7, with or without AV conduction defects, 108900 |
| NKX2-6 | 100.0% | 100.0% | 100.0% | 99.7% | Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095 |
| NODAL | 100.0% | 100.0% | 100.0% | 99.1% | Heterotaxy, visceral, 5, 270100 |
| NONO | 100.0% | 99.5% | 98.0% | 71.3% | Intellectual developmental disorder, X-linked syndromic 34, 300967 |
| NOS1AP | 100.0% | 100.0% | 100.0% | 98.7% | Nephrotic syndrome, type 22, 619155 |
| NOTCH1 | 100.0% | 100.0% | 100.0% | 99.6% | Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730 |
| NOTCH2 | 100.0% | 100.0% | 100.0% | 99.5% | Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500 |
| NPPA | 100.0% | 100.0% | 100.0% | 98.8% | Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201 |

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| NPPB | 100.0% | 100.0% | 100.0% | 99.5% | |
| NR2F2 | 100.0% | 100.0% | 99.9% | 96.6% | 46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779 |
| NRAP | 100.0% | 100.0% | 100.0% | 99.1% | |
| NRAS | 100.0% | 100.0% | 100.0% | 99.7% | Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500 |
| NSD1 | 100.0% | 100.0% | 100.0% | 98.6% | Sotos syndrome, 117550 |
| NUP155 | 100.0% | 100.0% | 100.0% | 97.8% | ?Atrial fibrillation 15, 615770 |
| PCCA | 100.0% | 100.0% | 100.0% | 98.4% | Propionicacidemia, 606054 |
| PCCB | 99.9% | 98.0% | 100.0% | 97.9% | Propionicacidemia, 606054 |
| PDLIM3 | 100.0% | 100.0% | 100.0% | 99.1% | |
| PDLIM5 | 99.7% | 97.8% | 100.0% | 98.9% | |

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| PEX5 | 100.0% | 100.0% | 100.0% | 98.8% | Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716 |
| PEX7 | 91.2% | 91.2% | 100.0% | 98.9% | Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879 |
| PGM1 | 94.0% | 94.0% | 100.0% | 98.0% | Congenital disorder of glycosylation, type It, 614921 |
| PHKA1 | 100.0% | 100.0% | 97.6% | 71.6% | Muscle glycogenosis, 300559 |
| PHYH | 100.0% | 100.0% | 100.0% | 98.2% | Refsum disease, 266500 |
| PITX2 | 100.0% | 100.0% | 100.0% | 98.1% | Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600 |
| PKD1L1 | 100.0% | 100.0% | 100.0% | 98.8% | Heterotaxy, visceral, 8, autosomal, 617205 |
| PKP2 | 99.9% | 99.3% | 99.9% | 98.0% | Arrhythmogenic right ventricular dysplasia 9, 609040 |
| PLD1 | 100.0% | 100.0% | 100.0% | 98.8% | Cardiac valvular dysplasia 1, 212093 |

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|---------|--------|--------|--------|-------|--|
| PLEKHM2 | 100.0% | 100.0% | 99.8% | 97.7% | |
| PLN | 100.0% | 100.0% | 100.0% | 98.7% | Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874 |
| PLXND1 | 100.0% | 100.0% | 100.0% | 98.5% | Congenital heart defects, multiple types, 9, 620294 |
| PMM2 | 100.0% | 100.0% | 100.0% | 98.1% | Congenital disorder of glycosylation, type Ia, 212065 |
| PNPLA2 | 100.0% | 100.0% | 100.0% | 99.5% | Neutral lipid storage disease with myopathy, 610717 |
| POMT1 | 100.0% | 100.0% | 100.0% | 98.1% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 |

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| POMT2 | 100.0% | 100.0% | 100.0% | 96.3% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 |
| PPA2 | 100.0% | 99.9% | 100.0% | 96.7% | ?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222 |
| PPCDC | 100.0% | 100.0% | 100.0% | 99.0% | |
| PPCS | 100.0% | 100.0% | 100.0% | 98.6% | Cardiomyopathy, dilated, 2C, 618189 |
| PPP1R13L | 100.0% | 99.9% | 99.8% | 95.7% | Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519 |
| PRDM16 | 100.0% | 100.0% | 99.8% | 98.4% | Left ventricular noncompaction 8, 615373;Cardiomyopathy, dilated, 1LL, 615373 |
| PRDM6 | 100.0% | 100.0% | 100.0% | 95.6% | Patent ductus arteriosus 3, 617039 |

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| PRKAG2 | 100.0% | 100.0% | 100.0% | 96.5% | Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858 |
| PRKD1 | 100.0% | 100.0% | 99.9% | 95.0% | Congenital heart defects and ectodermal dysplasia, 617364 |
| PTPN11 | 100.0% | 100.0% | 100.0% | 98.2% | Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785 |
| QRSL1 | 100.0% | 100.0% | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 40, 618835 |
| RAF1 | 100.0% | 100.0% | 100.0% | 98.7% | Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554 |
| RANGRF | 100.0% | 100.0% | 100.0% | 97.4% | |
| RBCK1 | 100.0% | 100.0% | 99.9% | 97.7% | Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 |
| RBFOX2 | 100.0% | 100.0% | 100.0% | 96.5% | |
| RBM20 | 100.0% | 100.0% | 100.0% | 99.2% | Cardiomyopathy, dilated, 1DD, 613172 |

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| RIT1 | 100.0% | 100.0% | 100.0% | 99.7% | Noonan syndrome 8, 615355 |
| ROBO4 | 100.0% | 100.0% | 100.0% | 98.9% | Aortic valve disease 3, 618496 |
| RPL3L | 100.0% | 100.0% | 100.0% | 99.2% | Cardiomyopathy, dilated, 2D, 619371 |
| RPS6KB1 | 100.0% | 100.0% | 100.0% | 97.6% | |
| RRAD | 100.0% | 100.0% | 100.0% | 96.7% | |
| RRAGC | 100.0% | 100.0% | 100.0% | 98.4% | Long-Olsen-Distelmaier syndrome, 620609 |
| RYR2 | 100.0% | 100.0% | 100.0% | 98.3% | Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 |
| SCN10A | 100.0% | 100.0% | 100.0% | 98.8% | Episodic pain syndrome, familial, 2, 615551 |
| SCN1B | 100.0% | 100.0% | 100.0% | 97.9% | Generalized epilepsy with febrile seizures plus, type 1, 604233;Developmental and epileptic encephalopathy 52, 617350;Cardiac conduction defect, nonspecific, 612838;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838 |

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|-------|--------|--------|--------|-------|---|
| SCN2B | 100.0% | 100.0% | 100.0% | 98.5% | Atrial fibrillation, familial, 14, 615378 |
| SCN3B | 100.0% | 100.0% | 100.0% | 99.0% | Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120 |
| SCN4B | 100.0% | 100.0% | 100.0% | 98.0% | Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819 |
| SCN5A | 100.0% | 100.0% | 100.0% | 98.6% | 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 |
| SCO2 | 100.0% | 100.0% | 100.0% | 99.5% | Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377 |

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| SDHA | 100.0% | 100.0% | 100.0% | 99.7% | Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma /paraganglioma syndrome 5, 614165 |
| SGCA | 100.0% | 100.0% | 100.0% | 99.1% | Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 |
| SGCB | 100.0% | 100.0% | 100.0% | 97.5% | Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 |
| SGCD | 100.0% | 99.8% | 100.0% | 99.4% | Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 |
| SGCG | 100.0% | 100.0% | 100.0% | 99.4% | Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 |
| SHMT2 | 100.0% | 100.0% | 100.0% | 99.6% | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 |
| SHOC2 | 100.0% | 100.0% | 100.0% | 97.1% | Noonan syndrome-like with loose anagen hair 1, 607721 |

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| SHROOM3 | 100.0% | 100.0% | 100.0% | 99.5% | |
| SLC22A5 | 100.0% | 100.0% | 100.0% | 98.4% | Carnitine deficiency, systemic primary, 212140 |
| SLC25A20 | 100.0% | 100.0% | 100.0% | 99.2% | Carnitine-acylcarnitine translocase deficiency, 212138 |
| SLC25A4 | 100.0% | 100.0% | 100.0% | 98.5% | Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283;Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 |
| SLC30A5 | 100.0% | 100.0% | 100.0% | 97.2% | |
| SLC4A3 | 100.0% | 100.0% | 100.0% | 98.9% | Short QT syndrome 7, 620231 |
| SLC6A6 | 100.0% | 100.0% | 100.0% | 98.4% | Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 |
| SLMAP | 100.0% | 100.0% | 100.0% | 98.0% | |
| SMAD1 | 100.0% | 100.0% | 100.0% | 98.0% | |
| SMAD6 | 100.0% | 100.0% | 99.8% | 91.5% | Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439 |

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| SMARCA4 | 100.0% | 100.0% | 100.0% | 99.5% | Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792 |
| SNTA1 | 100.0% | 100.0% | 99.9% | 95.8% | Long QT syndrome 12, 612955 |
| SOD2 | 100.0% | 100.0% | 100.0% | 99.4% | {Microvascular complications of diabetes 6}, 612634 |
| SOS1 | 100.0% | 100.0% | 100.0% | 96.8% | Noonan syndrome 4, 610733;?Fibromatosis, gingival, 1, 135300 |
| SOX7 | 100.0% | 100.0% | 99.8% | 92.0% | |
| SRF | 100.0% | 100.0% | 100.0% | 95.7% | |
| SRI | 100.0% | 100.0% | 100.0% | 98.8% | |
| SURF1 | 100.0% | 100.0% | 100.0% | 98.7% | Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110 |
| TAB2 | 100.0% | 100.0% | 100.0% | 98.3% | Congenital heart defects, nonsyndromic, 2, 614980 |
| TAF1 | 100.0% | 99.9% | 97.3% | 69.0% | Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250 |
| TAF1A | 100.0% | 100.0% | 100.0% | 97.1% | |

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|--------|--------|--------|--------|-------|---|
| TAZ | 100.0% | 100.0% | 96.7% | 66.1% | Barth syndrome, 302060 |
| TBX1 | 97.7% | 95.5% | 99.5% | 83.7% | Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430 |
| TBX20 | 100.0% | 100.0% | 100.0% | 98.5% | Atrial septal defect 4, 611363 |
| TBX5 | 100.0% | 100.0% | 100.0% | 99.0% | Holt-Oram syndrome, 142900 |
| TCAP | 100.0% | 100.0% | 100.0% | 99.9% | Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 |
| TDGF1 | 100.0% | 100.0% | 100.0% | 98.8% | |
| TECRL | 100.0% | 100.0% | 100.0% | 97.8% | Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021 |
| TFAP2B | 100.0% | 100.0% | 100.0% | 98.3% | Patent ductus arteriosus 2, 617035;Char syndrome, 169100 |
| TGFB3 | 100.0% | 100.0% | 100.0% | 99.5% | Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582 |
| THBS4 | 100.0% | 100.0% | 100.0% | 99.0% | |
| TJP1 | 100.0% | 100.0% | 100.0% | 98.9% | |

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|---------|--------|--------|--------|-------|---|
| TLL1 | 99.4% | 98.9% | 100.0% | 98.6% | Atrial septal defect 6, 613087 |
| TMEM260 | 100.0% | 100.0% | 100.0% | 97.9% | Structural heart defects and renal anomalies syndrome, 617478 |
| TMEM43 | 100.0% | 100.0% | 100.0% | 98.7% | Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302 |
| TMPO | 100.0% | 100.0% | 100.0% | 98.1% | |
| TNNC1 | 100.0% | 100.0% | 100.0% | 98.2% | Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243 |
| TNNI3 | 100.0% | 100.0% | 100.0% | 97.6% | ?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286 |
| TNNI3K | 100.0% | 100.0% | 100.0% | 98.6% | Cardiac conduction disease with or without dilated cardiomyopathy, 616117 |

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|----------|--------|--------|--------|-------|--|
| TNNT2 | 100.0% | 100.0% | 100.0% | 98.9% | Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494 |
| TNS1 | 100.0% | 100.0% | 100.0% | 99.0% | |
| TOR1AIP1 | 100.0% | 100.0% | 100.0% | 96.2% | ?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 |
| TPM1 | 100.0% | 100.0% | 100.0% | 98.0% | Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878 |
| TRDN | 99.9% | 99.6% | 100.0% | 96.8% | Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441 |
| TRIM63 | 100.0% | 100.0% | 100.0% | 98.2% | |
| TRPM4 | 100.0% | 100.0% | 100.0% | 98.9% | Progressive familial heart block, type IB, 604559;Erythrokeratoderma variabilis et progressiva 6, 618531 |

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|-------|--------|--------|--------|-------|---|
| TSC1 | 100.0% | 100.0% | 100.0% | 98.8% | Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangi leiomyomatosis, 606690 |
| TSFM | 94.3% | 94.3% | 100.0% | 98.5% | Combined oxidative phosphorylation deficiency 3, 610505 |
| TTN | 99.6% | 99.1% | 100.0% | 98.6% | Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Cardiomyopathy, familial hypertrophic, 9, 613765;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;Myopathy, myofibrillar, 9, with early respiratory failure, 603689 |
| TTR | 90.7% | 90.7% | 100.0% | 99.5% | Amyloidosis, hereditary, transthyretin-related, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680 |
| TULP3 | 100.0% | 100.0% | 100.0% | 99.2% | Hepatorenocardiac degenerative fibrosis, 619902 |

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|--------|--------|--------|--------|-------|---|
| TXNRD2 | 100.0% | 100.0% | 100.0% | 99.2% | ?Glucocorticoid deficiency 5, 617825 |
| VCL | 100.0% | 100.0% | 100.0% | 98.2% | Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255 |
| VEZF1 | 100.0% | 100.0% | 99.5% | 96.5% | ?Cardiomyopathy, dilated, 1OO, 620247 |
| XIRP2 | 100.0% | 100.0% | 100.0% | 97.5% | |
| XK | 100.0% | 99.9% | 97.9% | 71.5% | McLeod syndrome, 300842 |
| ZBTB17 | 100.0% | 100.0% | 100.0% | 99.7% | |
| ZFPM2 | 100.0% | 100.0% | 100.0% | 97.9% | Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500 |
| ZIC3 | 100.0% | 100.0% | 97.4% | 68.8% | Congenital heart defects, nonsyndromic, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390 |

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.

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.

srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.

This list is accurate for panel version DG 3.9.0

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