

HEART GENE PANEL DG 3.00 (306 genes)

Releasedate: 02-12-2020

| Gene | Agilent V5 covered > 10x | Agilent V5 covered > 20x | TWIST covered > 10x | TWIST covered 20x | Associated Phenotype description and OMIM disease ID |
|----------|--------------------------|--------------------------|---------------------|-------------------|---|
| AARS2 | 100 | 99,4 | 100 | 100 | Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889 |
| ABCC6 | 93,6 | 92,4 | 100 | 100 | Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473 |
| ABCC9 | 100 | 99,9 | 100 | 100 | Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 10, 608569 |
| ABL1 | 100 | 100 | 100 | 100 | Congenital heart defects and skeletal malformations syndrome, 617602 Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 |
| ACAD8 | 100 | 100 | 100 | 100 | Isobutyryl-CoA dehydrogenase deficiency, 611283 |
| ACAD9 | 100 | 99,9 | 100 | 100 | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| ACADVL | 99,4 | 97,3 | 100 | 100 | VLCAD deficiency, 201475 |
| ACSF3 | 100 | 99,9 | 100 | 100 | Combined malonic and methylmalonic aciduria, 614265 |
| ACTA2 | 100 | 99 | 100 | 100 | Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042 |
| ACTC1 | 100 | 99,7 | 100 | 100 | Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 |
| ACTN2 | 100 | 100 | 100 | 100 | Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654 |
| ACVR2B | 98,3 | 95 | 100 | 100 | Heterotaxy, visceral, 4, autosomal, 613751 |
| ADAMTS19 | 95 | 91,6 | 100 | 100 | No OMIM disease ID |

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|---------|------|------|------|------|--|
| ADCY5 | 95,1 | 91,2 | 99,2 | 98 | Dyskinesia, familial, with facial myokymia, 606703 |
| AGK | 90,6 | 88,6 | 91,2 | 91,2 | Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691 |
| AGL | 100 | 99,4 | 100 | 100 | Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400 |
| AGPAT2 | 99,6 | 96,1 | 100 | 100 | Lipodystrophy, congenital generalized, type 1, 608594 |
| AKAP9 | 98,8 | 95,5 | 100 | 100 | ?Long QT syndrome 11, 611820 |
| ALDH1A2 | 99,9 | 98,5 | 100 | 100 | No OMIM disease ID |
| ALMS1 | 99,8 | 99,5 | 100 | 100 | Alstrom syndrome, 203800 |
| ALPK3 | 97,8 | 94,6 | 100 | 100 | Cardiomyopathy, familial hypertrophic 27, 618052 |
| ANK2 | 100 | 100 | 100 | 100 | Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919 |
| ANKRD1 | 100 | 99,4 | 100 | 100 | No OMIM disease ID |
| ARIH1 | 100 | 99,5 | 100 | 100 | No OMIM disease ID |
| ATPAF2 | 100 | 100 | 100 | 100 | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 |
| BAG3 | 100 | 100 | 100 | 100 | Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954 |
| BANF1 | 98,3 | 86,6 | 100 | 100 | Nestor-Guillermo progeria syndrome, 614008 |
| BGN | 100 | 100 | 100 | 100 | Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106 |
| BMPR2 | 99,9 | 99,9 | 99,9 | 99,9 | Pulmonary venoocclusive disease 1, 265450 Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 |
| BRAF | 91 | 81,1 | 100 | 100 | Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0 |
| BSCL2 | 100 | 100 | 100 | 100 | Neuropathy, distal hereditary motor, type VC, 619112 Lipodystrophy, congenital generalized, type 2, 269700 |

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|----------|------|------|-----|-----|---|
| | | | | | Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924 |
| BVES | 99,9 | 98,8 | 100 | 100 | Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 |
| CACNA1C | 99,9 | 99,2 | 100 | 100 | Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875 |
| CACNA1D | 98 | 97,9 | 100 | 100 | Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896 |
| CACNA2D1 | 98,6 | 95,3 | 100 | 100 | No OMIM disease ID |
| CACNB2 | 98,6 | 98,5 | 100 | 100 | Brugada syndrome 4, 611876 |
| CALM1 | 100 | 99,4 | 100 | 100 | Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 |
| CALM2 | 67,8 | 65,1 | 72 | 72 | Long QT syndrome 15, 616249 |
| CALM3 | 100 | 99,1 | 100 | 100 | Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782 |
| CASQ2 | 100 | 100 | 100 | 100 | Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 |
| CAV1 | 100 | 100 | 100 | 100 | Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 |
| CAV3 | 100 | 100 | 100 | 100 | Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072 Cardiomyopathy, familial hypertrophic, 192600 |
| CDH2 | 99,3 | 97,7 | 100 | 100 | Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 |
| CFAP53 | 99,6 | 97,4 | 100 | 100 | Heterotaxy, visceral, 6, autosomal recessive, 614779 |
| CFC1 | 84,2 | 74,1 | 100 | 100 | Heterotaxy, visceral, 2, autosomal, 605376 |
| CHD7 | 100 | 99,5 | 100 | 100 | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800 |
| CHKB | 100 | 99,7 | 100 | 100 | Muscular dystrophy, congenital, megaconial type, 602541 |
| CHRM2 | 100 | 99,9 | 100 | 100 | No OMIM disease ID |

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| CITED2 | 99,2 | 99 | 100 | 100 | Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431 |
| COL3A1 | 99,6 | 97,6 | 100 | 100 | Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343 |
| COQ2 | 98 | 95,3 | 97,2 | 97,2 | {Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426 |
| COX15 | 99,9 | 98,8 | 100 | 100 | Mitochondrial complex IV deficiency, nuclear type 6, 615119 |
| CPT1A | 100 | 98,9 | 100 | 100 | CPT deficiency, hepatic, type IA, 255120 |
| CPT2 | 98,2 | 97,8 | 100 | 100 | CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 |
| CRELD1 | 99,9 | 95 | 100 | 100 | Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217 |
| CRYAB | 100 | 99,2 | 100 | 100 | Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Cataract 16, multiple types, 613763 |
| CSRP3 | 100 | 99,1 | 100 | 100 | Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482 |
| CTNNA3 | 100 | 99,8 | 100 | 100 | Arrhythmogenic right ventricular dysplasia, familial, 13, 615616 |
| DCHS1 | 99,8 | 99,1 | 100 | 100 | Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390 |
| DES | 100 | 99,7 | 100 | 100 | Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419 |
| DMD | 99,6 | 98,6 | 100 | 100 | Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200 |
| DOLK | 100 | 100 | 100 | 100 | Congenital disorder of glycosylation, type Im, 610768 |
| DPM3 | 100 | 100 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 |
| DPP6 | 99,7 | 97,8 | 99,4 | 97,6 | Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956 |

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| DSC2 | 99,8 | 98,4 | 100 | 100 | Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476 |
| DSG2 | 100 | 99,6 | 100 | 100 | Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877 |
| DSP | 100 | 99,6 | 100 | 100 | Keratosis palmoplantaris striata II, 612908 Arrhythmogenic right ventricular dysplasia 8, 607450 Epidermolysis bullosa, lethal acantholytic, 609638 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Skin fragility-woolly hair syndrome, 607655 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 |
| DTNA | 100 | 99,9 | 100 | 100 | Left ventricular noncompaction 1, with or without congenital heart defects, 604169 |
| DZIP1 | 98,4 | 96,6 | 100 | 100 | ?Mitral valve prolapse 3, 610840 Spermatogenic failure 47, 619102 |
| EEF1A2 | 100 | 100 | 99,9 | 99,1 | Mental retardation, autosomal dominant 38, 616393 Developmental and epileptic encephalopathy 33, 616409 |
| EFEMP2 | 100 | 100 | 100 | 100 | Cutis laxa, autosomal recessive, type IB, 614437 |
| EHMT1 | 94,5 | 93,7 | 99,6 | 99,5 | Kleefstra syndrome 1, 610253 |
| ELN | 99,8 | 97,8 | 100 | 100 | Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500 |
| EMD | 99,9 | 98,4 | 100 | 99,1 | Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 |
| EMILIN1 | 99,3 | 89,8 | 100 | 100 | No OMIM disease ID |
| ENPP1 | 96,4 | 91,2 | 98,7 | 97,8 | Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 |
| FAH | 100 | 100 | 100 | 100 | Tyrosinemia, type I, 276700 |
| FBN1 | 100 | 99,9 | 100 | 100 | Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900 |

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| FBN2 | 100 | 99,9 | 100 | 100 | Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118 |
| FBXO32 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| FGF12 | 99,9 | 98,1 | 100 | 100 | Developmental and epileptic encephalopathy 47, 617166 |
| FHL1 | 99,7 | 95,8 | 100 | 100 | Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696 |
| FHL2 | 99,9 | 98,7 | 100 | 100 | No OMIM disease ID |
| FHOD3 | 100 | 99,6 | 100 | 100 | No OMIM disease ID |
| FKRP | 100 | 100 | 100 | 99,9 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 |
| FKTN | 99,7 | 97 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 |
| FLNA | 100 | 99,9 | 100 | 100 | Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620 |
| FLNC | 100 | 99,6 | 100 | 100 | Cardiomyopathy, familial hypertrophic, 26, 617047 Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 |
| FLT4 | 99,2 | 98,3 | 100 | 100 | Congenital heart defects, multiple types, 7, 618780 Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 |

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| FNIP1 | 100 | 99,8 | 100 | 100 | No OMIM disease ID |
| FOXC2 | 100 | 96,7 | 100 | 99,8 | Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 |
| FOXD4 | 26,3 | 13,3 | 100 | 100 | No OMIM disease ID |
| FOXE3 | 82,6 | 72 | 94,4 | 87,8 | Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 |
| FOXH1 | 100 | 96,5 | 100 | 100 | No OMIM disease ID |
| FOXL1 | 96,6 | 89 | 100 | 100 | No OMIM disease ID |
| GAA | 100 | 99,9 | 100 | 100 | Glycogen storage disease II, 232300 |
| GATA4 | 84,1 | 74,5 | 100 | 99,9 | ?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 |
| GATA5 | 99,7 | 93,7 | 100 | 100 | Congenital heart defects, multiple types, 5, 617912 |
| GATA6 | 89,8 | 83 | 99,6 | 98 | Pancreatic agenesis and congenital heart defects, 600001 Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500 |
| GATAD1 | 99,9 | 97,9 | 100 | 99,1 | ?Cardiomyopathy, dilated, 2B, 614672 |
| GATB | 100 | 99,7 | 100 | 100 | ?Combined oxidative phosphorylation deficiency 41, 618838 |
| GATC | 100 | 100 | 100 | 100 | Combined oxidative phosphorylation deficiency 42, 618839 |
| GBE1 | 100 | 99,6 | 100 | 100 | Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500 |
| GDF1 | 73,9 | 54 | 98,7 | 92 | Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854 |
| GDF2 | 100 | 100 | 100 | 100 | Telangiectasia, hereditary hemorrhagic, type 5, 615506 |
| GJA1 | 100 | 100 | 100 | 100 | Erythrokeratoderma variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 |

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| | | | | | Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 |
| GJA5 | 100 | 100 | 100 | 100 | Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770 |
| GLA | 91,1 | 88,2 | 91,3 | 91,3 | Fabry disease, 301500 Fabry disease, cardiac variant, 301500 |
| GLB1 | 99,9 | 97,4 | 100 | 100 | GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600 |
| GMPPB | 100 | 100 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 |
| GNB2 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| GNPTAB | 100 | 99,9 | 100 | 100 | Mucopolipidosis II alpha/beta, 252500 Mucopolipidosis III alpha/beta, 252600 |
| GPD1L | 100 | 99,8 | 100 | 100 | Brugada syndrome 2, 611777 |
| HADHA | 97,2 | 91,6 | 100 | 100 | LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016 |
| HADHB | 98,8 | 89,7 | 100 | 100 | Trifunctional protein deficiency, 609015 |
| HAND1 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| HAND2 | 99,8 | 92,6 | 100 | 100 | No OMIM disease ID |
| HCN2 | 59,2 | 49,5 | 84,1 | 77,3 | No OMIM disease ID |
| HCN3 | 99,9 | 98,5 | 100 | 100 | No OMIM disease ID |
| HCN4 | 100 | 99,3 | 100 | 99,9 | Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800 |
| HEY2 | 100 | 99,3 | 100 | 100 | No OMIM disease ID |
| HFE | 100 | 99,7 | 100 | 100 | {Porphyria variegata, susceptibility to}, 176200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 |

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|--------|------|------|-----|-----|--|
| | | | | | [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200 |
| HJV | 100 | 100 | 100 | 100 | Hemochromatosis, type 2A, 602390 |
| HSPB6 | 91,1 | 81 | 100 | 100 | No OMIM disease ID |
| IDUA | 93,7 | 86,8 | 100 | 100 | Mucopolysaccharidosis lh/s, 607015 Mucopolysaccharidosis lh, 607014 Mucopolysaccharidosis ls, 607016 |
| ILK | 100 | 100 | 100 | 100 | No OMIM disease ID |
| ITPA | 100 | 100 | 100 | 100 | Developmental and epileptic encephalopathy 35, 616647 [Inosine triphosphatase deficiency], 613850 |
| JAG1 | 97,7 | 96,8 | 100 | 100 | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 |
| JPH2 | 95,5 | 80,3 | 100 | 100 | Cardiomyopathy, hypertrophic, 17, 613873 |
| JUP | 100 | 99,5 | 100 | 100 | Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214 |
| KCNA5 | 100 | 98,5 | 100 | 100 | Atrial fibrillation, familial, 7, 612240 |
| KCND2 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| KCND3 | 100 | 99,4 | 100 | 100 | Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346 |
| KCNE1 | 100 | 100 | 100 | 100 | Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347 |
| KCNE2 | 100 | 97,2 | 100 | 100 | Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693 |
| KCNE3 | 100 | 100 | 100 | 100 | ?Brugada syndrome 6, 613119 |
| KCNE4 | 80,5 | 80,4 | 100 | 100 | No OMIM disease ID |
| KCNE5 | 98,6 | 91,8 | 100 | 100 | No OMIM disease ID |
| KCNH2 | 95,8 | 91,9 | 100 | 100 | {Long QT syndrome 2, acquired, susceptibility to}, 613688 Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 |
| KCNJ11 | 100 | 100 | 100 | 100 | Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853 |

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|--------|------|------|-----|------|---|
| | | | | | Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820 |
| KCNJ2 | 100 | 100 | 100 | 100 | Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 |
| KCNJ5 | 100 | 100 | 100 | 100 | Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677 |
| KCNJ8 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| KCNK3 | 97,5 | 95 | 100 | 100 | Pulmonary hypertension, primary, 4, 615344 |
| KCNN3 | 100 | 99,7 | 100 | 100 | Zimmermann-Laband syndrome 3, 618658 |
| KCNQ1 | 93,3 | 90,6 | 100 | 99,8 | Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500 Atrial fibrillation, familial, 3, 607554 |
| KLF10 | 100 | 99,9 | 100 | 100 | No OMIM disease ID |
| KLHL24 | 100 | 100 | 100 | 100 | Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294 |
| KMT2D | 100 | 99,4 | 100 | 100 | Kabuki syndrome 1, 147920 |
| KRAS | 99,5 | 96,9 | 100 | 100 | Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942 |
| LAMA2 | 100 | 99,6 | 100 | 100 | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 |
| LAMA4 | 100 | 99,9 | 100 | 100 | Cardiomyopathy, dilated, 1JJ, 615235 |

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| LAMP2 | 99,2 | 95,6 | 100 | 100 | Danon disease, 300257 |
| LDB3 | 95,4 | 94,7 | 100 | 100 | Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493 |
| LEFTY2 | 88,9 | 81,4 | 100 | 100 | No OMIM disease ID |
| LIMS2 | 93 | 92,7 | 99,8 | 98,9 | ?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 |
| LMNA | 97,4 | 91,9 | 100 | 100 | Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112 |
| LMOD1 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| LOX | 100 | 99,6 | 100 | 100 | Aortic aneurysm, familial thoracic 10, 617168 |
| LRRC10 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| LTBP3 | 99,6 | 98,1 | 100 | 100 | Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809 |
| LZTR1 | 100 | 99,9 | 100 | 100 | {Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564 |
| MAT2A | 99,6 | 96,4 | 100 | 100 | No OMIM disease ID |
| MCTP2 | 99,7 | 98,2 | 100 | 100 | No OMIM disease ID |
| MED13L | 100 | 99,8 | 100 | 100 | Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789 |
| MFAP5 | 99,9 | 97,6 | 100 | 100 | Aortic aneurysm, familial thoracic 9, 616166 |
| MIB1 | 100 | 99,9 | 100 | 100 | Left ventricular noncompaction 7, 615092 |

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|--------|------|------|-----|------|--|
| MLYCD | 96 | 90,4 | 100 | 98,9 | Malonyl-CoA decarboxylase deficiency, 248360 |
| MMP21 | 99,9 | 98,8 | 100 | 100 | Heterotaxy, visceral, 7, autosomal, 616749 |
| MYBPC3 | 99,9 | 97,6 | 100 | 100 | Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396 |
| MYBPHL | 99,9 | 98,6 | 100 | 100 | No OMIM disease ID |
| MYH11 | 100 | 100 | 100 | 100 | Aortic aneurysm, familial thoracic 4, 132900 |
| MYH6 | 99,4 | 97,1 | 100 | 100 | Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090 Cardiomyopathy, dilated, 1EE, 613252 |
| MYH7 | 99,6 | 97,3 | 100 | 100 | Myopathy, myosin storage, autosomal recessive, 255160 Left ventricular noncompaction 5, 613426 Laing distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Cardiomyopathy, hypertrophic, 1, 192600 |
| MYH7B | 98,4 | 94,2 | 100 | 100 | No OMIM disease ID |
| MYL2 | 96,1 | 88,1 | 99 | 95,4 | Cardiomyopathy, hypertrophic, 10, 608758 |
| MYL3 | 100 | 100 | 100 | 100 | Cardiomyopathy, hypertrophic, 8, 608751 |
| MYL4 | 100 | 100 | 100 | 100 | ?Atrial fibrillation, familial, 18, 617280 |
| MYL7 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| MYLK | 100 | 99,9 | 100 | 100 | Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210 |
| MYLK2 | 100 | 100 | 100 | 100 | Cardiomyopathy, hypertrophic, 1, digenic, 192600 |
| MYLK3 | 99,4 | 97,8 | 100 | 100 | No OMIM disease ID |
| MYO6 | 99,5 | 96,6 | 100 | 100 | Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 |
| MYOM1 | 99,9 | 98,4 | 100 | 100 | No OMIM disease ID |

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|--------|------|------|------|------|--|
| MYOT | 100 | 99,6 | 100 | 100 | Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920 |
| MYOZ2 | 100 | 100 | 100 | 100 | Cardiomyopathy, hypertrophic, 16, 613838 |
| MYPN | 100 | 99,7 | 100 | 100 | Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248 |
| MYRF | 99,3 | 98,5 | 100 | 100 | Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 |
| NEBL | 99,2 | 97,1 | 100 | 100 | No OMIM disease ID |
| NEXN | 92 | 77,5 | 100 | 99,9 | Cardiomyopathy, hypertrophic, 20, 613876 Cardiomyopathy, dilated, 1CC, 613122 |
| NKX2-5 | 100 | 99,7 | 100 | 100 | Ventricular septal defect 3, 614432 Tetralogy of Fallot, 187500 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095 Hypothyroidism, congenital nongoitrous, 5, 225250 Atrial septal defect 7, with or without AV conduction defects, 108900 |
| NKX2-6 | 100 | 99,5 | 100 | 100 | Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095 |
| NNT | 96,4 | 95,9 | 96,4 | 96,4 | Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 |
| NODAL | 100 | 100 | 100 | 100 | Heterotaxy, visceral, 5, 270100 |
| NOS1AP | 100 | 99,9 | 100 | 100 | No OMIM disease ID |
| NOTCH1 | 99,2 | 97,2 | 100 | 100 | Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028 |
| NOTCH2 | 100 | 99,5 | 100 | 100 | Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205 |
| NPPA | 100 | 100 | 100 | 100 | Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201 |
| NPPB | 100 | 100 | 100 | 100 | No OMIM disease ID |
| NR2F2 | 100 | 98,5 | 100 | 100 | Congenital heart defects, multiple types, 4, 615779 46,XX sex reversal 5, 618901 |
| NRAS | 100 | 100 | 100 | 100 | Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 |

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|---------|------|------|------|------|--|
| | | | | | Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 |
| NUP155 | 99,2 | 97,4 | 100 | 100 | ?Atrial fibrillation 15, 615770 |
| OBSCN | 99,3 | 98,1 | 100 | 99,9 | No OMIM disease ID |
| PCCA | 99,5 | 96,7 | 100 | 100 | Propionicacidemia, 606054 |
| PCCB | 97,9 | 96 | 98,7 | 96,2 | Propionicacidemia, 606054 |
| PDLIM3 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |
| PDLIM5 | 93,5 | 91,3 | 97,5 | 95,2 | No OMIM disease ID |
| PEX5 | 99,9 | 99 | 100 | 100 | Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110 |
| PEX7 | 87,8 | 80,7 | 91,3 | 91,3 | Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100 |
| PGM1 | 94,2 | 94,2 | 94,2 | 94,2 | Congenital disorder of glycosylation, type It, 614921 |
| PHKA1 | 99,2 | 95,3 | 100 | 99,9 | Muscle glycogenosis, 300559 |
| PHYH | 100 | 99,6 | 100 | 100 | Refsum disease, 266500 |
| PITX2 | 99,9 | 97,7 | 100 | 100 | Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600 |
| PKD1L1 | 100 | 99,8 | 100 | 100 | Heterotaxy, visceral, 8, autosomal, 617205 |
| PKP2 | 95,4 | 88,6 | 95 | 95 | Arrhythmogenic right ventricular dysplasia 9, 609040 |
| PKP4 | 99,8 | 98,2 | 100 | 100 | No OMIM disease ID |
| PLD1 | 100 | 99,6 | 100 | 100 | Cardiac valvular defect, developmental, 212093 |
| PLEKHM2 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| PLN | 100 | 100 | 100 | 100 | Cardiomyopathy, hypertrophic, 18, 613874 Cardiomyopathy, dilated, 1P, 609909 |
| PLOD1 | 100 | 98,4 | 100 | 100 | Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 |

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|--------|------|------|------|------|---|
| PMEP1 | 100 | 99,2 | 100 | 99,9 | No OMIM disease ID |
| PMM2 | 100 | 100 | 100 | 100 | Congenital disorder of glycosylation, type Ia, 212065 |
| PNPLA2 | 99,7 | 96,1 | 100 | 100 | Neutral lipid storage disease with myopathy, 610717 |
| POMT1 | 99,3 | 97,5 | 100 | 100 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 |
| POMT2 | 99,4 | 96,4 | 100 | 100 | 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 mental retardation), type B, 2, 613156 |
| PPA2 | 98,7 | 94 | 100 | 100 | ?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222 |
| PPCS | 99,8 | 99,5 | 100 | 100 | Cardiomyopathy, dilated, 2C, 618189 |
| PRDM16 | 99,8 | 99,1 | 100 | 100 | Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373 |
| PRKAG2 | 99,1 | 96,7 | 100 | 99,4 | Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858 |
| PRKG1 | 92,5 | 91,2 | 92,7 | 92,7 | Aortic aneurysm, familial thoracic 8, 615436 |
| PTPN11 | 99,1 | 93,7 | 100 | 100 | LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785 |
| QRSL1 | 99,2 | 93,9 | 100 | 100 | Combined oxidative phosphorylation deficiency 40, 618835 |
| RAF1 | 100 | 100 | 100 | 100 | LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916 |
| RANGRF | 100 | 99,9 | 100 | 100 | No OMIM disease ID |
| RBM20 | 100 | 99,9 | 100 | 100 | Cardiomyopathy, dilated, 1DD, 613172 |
| RIT1 | 100 | 100 | 100 | 100 | Noonan syndrome 8, 615355 |
| RRAD | 85,4 | 80,7 | 99,4 | 96,2 | No OMIM disease ID |
| RRAGC | 100 | 99,7 | 100 | 100 | No OMIM disease ID |

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|---------|------|------|------|------|---|
| RYR2 | 99,9 | 99 | 100 | 100 | Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996 |
| SCN10A | 100 | 99,6 | 100 | 100 | Episodic pain syndrome, familial, 2, 615551 |
| SCN1B | 98 | 96,4 | 99,8 | 99,3 | Atrial fibrillation, familial, 13, 615377 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838 |
| SCN2B | 100 | 100 | 100 | 100 | Atrial fibrillation, familial, 14, 615378 |
| SCN3B | 100 | 100 | 100 | 100 | Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120 |
| SCN4B | 100 | 99,6 | 100 | 100 | Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819 |
| SCN5A | 99 | 99 | 100 | 100 | Atrial fibrillation, familial, 10, 614022 Sick sinus syndrome 1, 608567 Long QT syndrome 3, 603830 Heart block, nonprogressive, 113900 Cardiomyopathy, dilated, 1E, 601154 Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900 {Sudden infant death syndrome, susceptibility to}, 272120 Ventricular fibrillation, familial, 1, 603829 |
| SDHA | 85,8 | 80,4 | 100 | 100 | Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Parangliomas 5, 614165 Mitochondrial respiratory chain complex II deficiency, 252011 |
| SGCA | 100 | 99,9 | 100 | 100 | Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 |
| SGCB | 97,7 | 96,5 | 100 | 100 | Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 |
| SGCD | 100 | 98,9 | 100 | 100 | Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 |
| SGCG | 100 | 99,2 | 100 | 100 | Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 |
| SHOC2 | 99,9 | 99,4 | 100 | 100 | Noonan syndrome-like with loose anagen hair 1, 607721 |
| SHROOM3 | 98,6 | 97,8 | 100 | 100 | No OMIM disease ID |
| SKI | 99,3 | 94,9 | 100 | 99,4 | Shprintzen-Goldberg syndrome, 182212 |

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|----------|------|------|------|------|---|
| SLC22A5 | 100 | 100 | 100 | 100 | Carnitine deficiency, systemic primary, 212140 |
| SLC25A20 | 100 | 100 | 100 | 100 | Carnitine-acylcarnitine translocase deficiency, 212138 |
| SLC25A4 | 100 | 100 | 100 | 100 | Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 |
| SLC2A10 | 97,7 | 97,7 | 100 | 100 | Arterial tortuosity syndrome, 208050 |
| SLMAP | 99,2 | 94,6 | 100 | 100 | No OMIM disease ID |
| SMAD1 | 100 | 99,1 | 100 | 100 | No OMIM disease ID |
| SMAD2 | 100 | 99,6 | 100 | 100 | No OMIM disease ID |
| SMAD3 | 99,9 | 99 | 100 | 100 | Loeys-Dietz syndrome 3, 613795 |
| SMAD4 | 100 | 99,9 | 100 | 100 | Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 |
| SMAD6 | 90,9 | 81 | 100 | 99,6 | Aortic valve disease 2, 614823 {Radioulnar synostosis, nonsyndromic}, 179300 {Craniosynostosis 7, susceptibility to}, 617439 |
| SMAD9 | 100 | 99,9 | 100 | 100 | Pulmonary hypertension, primary, 2, 615342 |
| SNTA1 | 87 | 78,8 | 99,3 | 97,2 | Long QT syndrome 12, 612955 |
| SOS1 | 99,8 | 98,4 | 100 | 100 | Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300 |
| SRI | 99,9 | 97,8 | 100 | 100 | No OMIM disease ID |
| SYNE1 | 98,2 | 97,8 | 98,8 | 98,8 | Arthrogryposis multiplex congenita 3, myogenic type, 618484 Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 |
| SYNE2 | 99,7 | 98,1 | 100 | 99,9 | Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 |
| TAB2 | 100 | 99,7 | 100 | 100 | Congenital heart defects, nonsyndromic, 2, 614980 |
| TAZ | 99,1 | 95,5 | 100 | 100 | Barth syndrome, 302060 |
| TBX1 | 87 | 77,5 | 94 | 89,9 | Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 |

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|--------|------|------|-----|------|--|
| | | | | | DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 |
| TBX20 | 100 | 99,7 | 100 | 100 | Atrial septal defect 4, 611363 |
| TBX5 | 100 | 100 | 100 | 100 | Holt-Oram syndrome, 142900 |
| TCAP | 100 | 100 | 100 | 100 | Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 |
| TDGF1 | 99,9 | 96,7 | 100 | 100 | Forebrain defects, 0 |
| TECRL | 96,3 | 89,3 | 100 | 100 | Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021 |
| TFAP2B | 99,9 | 98,6 | 100 | 100 | Char syndrome, 169100 Patent ductus arteriosus 2, 617035 |
| TGFB2 | 100 | 100 | 100 | 100 | Loeys-Dietz syndrome 4, 614816 |
| TGFB3 | 100 | 100 | 100 | 100 | Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970 |
| TGFBR1 | 93,7 | 93,6 | 99 | 96,3 | Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 |
| TGFBR2 | 100 | 100 | 100 | 100 | Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168 |
| THBS4 | 99,9 | 99,4 | 100 | 100 | No OMIM disease ID |
| TJP1 | 100 | 99,7 | 100 | 100 | No OMIM disease ID |
| TLL1 | 100 | 100 | 100 | 100 | Atrial septal defect 6, 613087 |
| TMEM43 | 99,9 | 98,9 | 100 | 100 | Emery-Dreifuss muscular dystrophy 7, AD, 614302 Arrhythmogenic right ventricular dysplasia 5, 604400 |
| TMPO | 98,4 | 94,7 | 100 | 100 | No OMIM disease ID |
| TNNC1 | 100 | 100 | 100 | 100 | Cardiomyopathy, hypertrophic, 13, 613243 Cardiomyopathy, dilated, 1Z, 611879 |
| TNNI3 | 99,7 | 95,4 | 100 | 100 | Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 |
| TNNI3K | 100 | 99,4 | 100 | 100 | Cardiac conduction disease with or without dilated cardiomyopathy, 616117 |
| TNNT2 | 94,8 | 91,1 | 100 | 99,3 | Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 |

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|----------|------|------|------|------|---|
| | | | | | Left ventricular noncompaction 6, 601494 Cardiomyopathy, dilated, 1D, 601494 |
| TOR1AIP1 | 99,9 | 98 | 100 | 100 | ?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 |
| TPM1 | 100 | 99,4 | 100 | 99,9 | Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878 |
| TRDN | 96,2 | 86,7 | 100 | 100 | Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441 |
| TRIM63 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| TRPM4 | 100 | 99,5 | 100 | 100 | Erythrokeratoderma variabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559 |
| TSMF | 100 | 99,5 | 94,9 | 94,9 | Combined oxidative phosphorylation deficiency 3, 610505 |
| TTN | 98,6 | 98,1 | 100 | 100 | Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 |
| TTR | 94,6 | 94,6 | 94,6 | 94,6 | Amyloidosis, hereditary, transthyretin-related, 105210 [Dystransthyretinemic hyperthyroxinemia], 145680 Carpal tunnel syndrome, familial, 115430 |
| TXNRD2 | 96,8 | 95,9 | 100 | 100 | ?Glucocorticoid deficiency 5, 617825 |
| VCL | 99,9 | 99 | 100 | 100 | Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255 |
| XIRP2 | 100 | 99,9 | 100 | 99,9 | No OMIM disease ID |
| XK | 99,8 | 98,1 | 100 | 100 | McLeod syndrome with or without chronic granulomatous disease, 300842 |
| ZBTB17 | 100 | 100 | 100 | 100 | No OMIM disease ID |
| ZFPM2 | 100 | 100 | 100 | 100 | 46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500 Diaphragmatic hernia 3, 610187 |
| ZIC3 | 100 | 99,9 | 100 | 100 | Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390 |

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.

Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.

TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.

non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.

This list is accurate for panel version DG 3.0.0

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