HEART GENE PANEL DG 2.14 (346 genes)

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCC6	116.4	93.6	92.6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	157.9	99.9	99.2	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAD8	141.5	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	135.2	98.4	95.7	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADVL	119	98.7	95.1	VLCAD deficiency, 201475
ACSF3	128.8	99.9	99.3	Combined malonic and methylmalonic aciduria, 614265
ACTA1	99.7	99.2	95.3	?Myopathy, scapulohumeroperoneal, 616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACTA2	137.6	100	99.8	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTC1	164.1	100	99.6	Atrial septal defect 5, 612794 cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTN1	143.6	100	99.9	Bleeding disorder, platelet-type, 15, 615193
ACTN2	156.3	100	100	cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACVR1	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100
ACVR2B	140.5	97.1	94.7	Heterotaxy, visceral, 4, autosomal, 613751
ADCY5	129.2	92.4	89.2	Dyskinesia, familial, with facial myokymia, 606703
ADRB1	155.1	97.2	89.8	[Resting heart rate], 607276

				{Congestive heart failure and beta-blocker response, modifier of}, 0
ADRB2	131.3	100	100	Beta-2-adrenoreceptor agonist, reduced response to, 0 {Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665
AGK	112.1	99.3	96.4	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146.7	99.7	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	109.5	99	95.1	Lipodystrophy, congenital generalized, type 1, 608594
AGRN	114.8	95.2	89.3	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	214.2	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTR1	134.6	92	91.9	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AKAP9	98	98.3	94.2	?Long QT syndrome-11, 611820
ALDH1A2	114.8	100	99.6	No OMIM phenotype Tetralogy of Fallot (Pavan (2009) BMC Med Genet 10, 113) Pentalogy of Cantrell (Steiner (2013) J Med Case Rep 7,287) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89,476)
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
ALPK3	98.7	94.6	92.5	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	160.3	100	99.9	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKRD1	101.7	99.5	96.8	No OMIM phenotype cardiomyopathy,hypertrophic (Arimura (2009) J Am Coll Cardiol 54,334) Cardiomyopathy,dilated (Duboscq-Bidot (2009) Eur Heart J 30,2128) ?Total anomalous pulmonary venous return (Cinquetti (2008) Hum Mutat 29,468) ?Neurodevelopmental disorder (Handrigan (2013) J Med Genet 50,163)
ATP1A4	161.6	100	99.5	No OMIM phenotype
ATPAF2	101.5	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	136.5	100	100	cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	58.3	98	88.1	Nestor-Guillermo progeria syndrome, 614008

BGN	128.9	100	99.5	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BRCC3	53	83.8	63.2	No OMIM phenotype
BSCL2	113.5	100	100	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BVES	114.8	99.8	98.2	?Muscular dystrophy, limb-girdle, type 2X, 616812
CACNA1C	154.6	99.9	99.2	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	149.9	98	97.8	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	82.6	93.1	84.4	No OMIM phenotype Brugada syndrome (Burashnikov (2010) Heart Rhythm 7,1872) Short QT syndrome (Templin (2011) Eur Heart J 32,1077) Histiocytoid Cardiomyopathy (Cataldo (2014) Cardiol Young epub) West syndrome (Hino-Fukuyo (2015) Hum Genet 134,649)
CACNA2D4	112	99.2	97.7	Retinal cone dystrophy 4, 610478
CACNB2	150.9	99.5	96.9	Brugada syndrome 4, 611876
CALM1	114.3	100	99.7	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	54	67.8	65.8	Long QT syndrome 15, 616249
CALM3	117.4	99.9	99.5	No OMIM phenotype Catecholaminergic polymorphic ventricular tachycardia (Boczek (2013) Circulation 128,A14699) Long QT syndrome (Reed (2015) Heart Rhythm 12,419) {Cardiomyopathy,hypertrophic,modifier of} (Friedrich (2009) Eur Heart J 30,1648)
CAMK2D	106.9	97.3	93.8	No OMIM phenotype
CARD6	149.6	97.5	97.1	No OMIM phenotype
CASQ2	143.3	99.9	99.2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV1	265.4	100	100	?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 Pulmonary hypertension, primary, 3, 615343
CAV3	304.7	100	100	cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320

				Long QT syndrome 9, 611818 Muscular dystrophy, limb-girdle, type IC, 607801 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072
CAVIN4	159.7	100	100	No OMIM phenotype
CDH2	137.5	98.5	97.5	No OMIM phenotype
CFAP53	146.6	97.6	94.2	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	74.5	82.7	71.3	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	142.9	100	99.8	No OMIM phenotype
CIB1	131.1	95.2	92.5	No OMIM phenotype
CITED2	111.6	99.2	99	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	104.3	97.8	92.3	Ehlers-Danlos syndrome, vascular type, 130050
COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COX15	98.6	100	99.7	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
CPT1A	169.3	100	98.7	CPT deficiency, hepatic, type IA, 255120
CPT2	162.8	97.2	95.4	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CRELD1	114.4	99.9	97.8	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRKL	166.4	100	99.8	No OMIM phenotype ?Congenital heart defect (Breckpot (2012) Am J Med Genet A 158A,574) ?Tetralogy of Fallot (Tomita-Mitchell (2012) Physiol Genomics 44,518) ?Ventricular septal defect (Zhao (2013) Am J Med Genet A 161,3087
CRYAB	125.7	99.9	98.7	cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810

				Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CSRP3	103	100	99.9	?cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CTF1	24.5	27.8	20	No OMIM phenotype Cardiomyopathy,dilated (Erdmann (2000) Hum Mutat 16,448)
CTNNA3	138.3	100	99.9	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CXADR	95	95.9	88.6	No OMIM phenotype
DES	120.8	99.9	98.1	?Muscular dystrophy, limb-girdle, type 2R, 615325 cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DMD	111.5	99.4	97.4	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMPK	117.7	99.9	97.9	Myotonic dystrophy 1, 160900
DNM2	127.4	97.5	94.4	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DOLK	202.9	100	99.9	Congenital disorder of glycosylation, type Im, 610768
DPM3	183.7	100	100	Congenital disorder of glycosylation, type Io, 612937
DPP6	145.5	96.5	94.5	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	128.5	99.4	96.2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSG2	140.6	99.9	98.7	Arrhythmogenic right ventricular dysplasia 10, 610193 cardiomyopathy, dilated, 1BB, 612877
DSP	154	100	99.8	Arrhythmogenic right ventricular dysplasia 8, 607450 cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DTNA	156.5	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169

EDN1	145.5	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EEF1A2	177.7	98.8	93.8	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP2	120.9	100	99.9	Cutis laxa, autosomal recessive, type IB, 614437
ELN	91.1	99.4	97.4	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
EMD	100.3	99.8	97.2	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMILIN1	75.6	96.8	87.5	No OMIM phenotype Connective tissue disease, autosomal dominant (Capuano (2016) Hum Mutat 37, 84)
ENG	128.8	97.4	93.6	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	134.8	92.4	83.2	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
EPG5	126	99.3	97.7	Vici syndrome, 242840
EYA4	160.6	100	99.5	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FAH	151.3	100	99.9	Tyrosinemia, type I, 276700
FBN1	159.8	99.9	99.5	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FBXO32	150.5	100	100	No OMIM phenotype
FGF12	95.5	99.6	96.3	Epileptic encephalopathy, early infantile, 47, 617166
FGF13	102.6	99.7	97.6	No OMIM phenotype
FHL1	87.2	98.8	93	?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696

				Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy, X-linked dominant, 300695
FHL2	149.6	99.6	98.4	No OMIM phenotype Cardiomyopathy,hypertrophic (Friedrich (2014) Basic Res Cardiol 109,451) ?Distal myopathy (Evila (2016) Neuromuscul Disord 26,7)
FHOD3	135.3	99.9	98.5	No OMIM phenotype
FKRP	94.5	100	99.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	120.2	99.2	94.2	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNA	138.1	100	99.5	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	150	99.8	99.2	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLNC	165.1	100	99.7	Cardiomyopathy, familial hypertrophic, 26, 0 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLT4	155.9	98.6	97.9	Hemangioma, capillary infantile, somatic, 602089

				Lymphedema, hereditary, IA, 153100
FOXD4	3.1	25.1	13.3	No OMIM phenotype
FOXE3	20.6	69	47.8	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXH1	47.2	98.5	85	No OMIM phenotype Congenital heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J cardiol 145,83)
FXN	75.4	85.9	75.9	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GAA	128.5	100	99.9	Glycogen storage disease II, 232300
GATA4	87.4	68.6	60.7	?Testicular anomalies with or without cardio, 615542 Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429
GATA5	44.2	98.3	84.4	Congenital heart defects, multiple types, 5, 617912
GATA6	61.7	83.7	72.1	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	126.8	97	92	?Cardiomyopathy, dilated, 2B, 614672
GBE1	145.5	99.6	97.2	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDF1	19.5	65	48.4	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF2	163.2	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GJA1	246.4	100	100	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100

				Syndactyly, type III, 186100
GJA5	268.4	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJC1	192.8	100	100	No OMIM phenotype
GLA	81.3	99.7	97.6	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GMPPB	228.5	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
GNB5	125.8	99.9	98.3	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	153.8	100	99.8	Nonaka myopathy, 605820 Sialuria, 269921
GNPTAB	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GPD1L	138.4	100	98.5	Brugada syndrome 2, 611777
H19				Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
HADHA	84.4	96.5	90.3	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	80.5	92.5	79.5	Trifunctional protein deficiency, 609015
HAND1	84.8	100	98.9	No OMIM phenotype Ventricular septal defect (Cheng (2011) Clin Chim Acta) Cardiac malformations (Reamon-Buettner (2009) Hum Mol Genet 18,3567) cardiomyopathy, dilated (Zhou (2015) Clin Chem Lab Med Epub, epub)
HAND2	32.2	87.8	67.9	No OMIM phenotype Tetralogy of Fallot (Topf (2014) PLoS One 9,e95453) Ventricular septal defect (Sun (2016) G3 (Bethesda) epub,epub)

				?cardio (Shen (2010) Chin Med J (Engl) 123,1623)
HCN1	122.4	99.9	97.8	Epileptic encephalopathy, early infantile, 24, 615871
HCN2	53.2	58.3	50.7	No OMIM phenotype Epilepsy, generalised (Li (2018) Hum Mutat 39,202) ?Tetralogy of Fallot (Grunert (2014) Hum Mol Genet 23, 3115)
HCN3	146.4	99.9	99.2	No OMIM phenotype
HCN4	79.4	98.3	91.8	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HEY2	146.1	99.2	92.8	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
HFE	142.3	100	99.7	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	117	100	100	Hemochromatosis, type 2A, 602390
HRAS	164.7	99.8	98.1	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HSPB6	63.1	89.4	77.4	No OMIM phenotype
IDUA	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
ILK	171.7	100	100	No OMIM phenotype Cardiomyopathy, dilated (Knoll (2007) Circulation 116,515) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89, 476)
IRX3	73.8	82.9	66.2	No OMIM phenotype
IRX4	90	95.8	92.3	No OMIM phenotype Congenital heart defect (Cheng (2014) BMC Genomics 15,1127) {Prostate cancer,susceptibility to} (Nguyen (2012) Hum Mol Genet 21,2076)

ITGB1BP2	74.6	99.3	95.7	No OMIM phenotype
ITPA	120.2	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAG1	148.4	98.1	97.5	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JPH2	87.5	90.4	75.1	cardiomyopathy, hypertrophic, 17, 613873
JUP	145.1	100	99.6	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KAT6B	192.3	99.6	98.5	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KCNA5	143.3	99.4	96	Atrial fibrillation, familial, 7, 612240
KCND2	183.4	100	100	No OMIM phenotype Autism and epilepsy (Lee (2014) Hum Mol Genet 23,3481) J-wave syndrome with sudden cardiac death (Perrin (2014) Circ Cardiovasc Genet 7,782) Epilepsy,temporal lobe (Singh (2006) Neurobiol Dis 24,245)
KCND3	182.5	99.9	99.1	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	462.6	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	181.9	100	100	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	176.8	100	100	Brugada syndrome 6, 613119
KCNE4	84.9	79.9	77.6	No OMIM phenotype ?Periodic paralysis (Silva (2004) Arq Bras Endocrinol Metabol 48,196) {Atrial fibrillation, association with} (Zeng (2007) Cardiology 108,97)
KCNE5	87.4	97.6	90.1	No OMIM phenotype Atrial fibrillation (Ravn (2008) Heart Rhythm 5,427 Idiopathic ventricular fibrillation (Ohno (2011) Circ Arrhythm Electrophysiol 4,352) Atrial fibrillation,lone,early-onset (Olesen (2014) Heart Rhythm 11,246)
KCNH2	102.8	92.3	84.8	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ11	299.5	100	100	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176

				Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ12	575.4	100	100	No OMIM phenotype
KCNJ2	229.1	100	100	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ3	168.1	100	100	No OMIM phenotype {Schizophrenia, association with} (Yamada (2012) Hum Genet 131,443)
KCNJ5	194.1	100	99.8	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ8	177.3	100	100	No OMIM phenotype Cantu syndrome (Brownstein (2013) Eur J Med Genet 56,678) Sudden infant death syndrome (Klaver (2011) Int J Cardiol 152,162) ?Ventricular fibrillation (Haissaguerre (2009) J Cardiovasc Electrophysiol 20,93)
KCNK3	165.4	98.9	96.1	Pulmonary hypertension, primary, 4, 615344
KCNMB1	116	100	100	{Hypertension, diastolic, resistance to}, 608622
KCNN2	169.8	99.5	99.5	No OMIM phenotype
KCNN3	151.3	100	99.9	No OMIM phenotype Non-cirrhotic portal hypertension (Koot (2016) J Hepatol 64,974)
KCNQ1	114.7	93	90.3	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ1OT1				Beckwith-Wiedemann syndrome, 130650
KLF10	141.4	100	99.7	No OMIM phenotype
KLHL24	192.9	100	100	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KRAS	64.7	99.9	98.7	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980

				Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LAMA2	143.5	99.9	99.5	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMA4	132.6	100	99.9	cardiomyopathy, dilated, 1JJ, 615235
LAMP2	106.1	92.7	91.2	Danon disease, 300257
LDB3	127.3	95.5	93.7	cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LEFTY2	42.3	91.3	77.1	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LIMS2	110.8	93	92.3	Muscular dystrophy, limb-girdle, type 2W, 616827
LMNA	89.2	97.9	91.3	cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, AD, 181350 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210
LMOD1	177.4	100	100	No OMIM phenotype Megacystis-microcolon-intestinal hypoperistalsis syndrome (Halim (2017) Proc Natl Acad Sci USA 114)
LOX	104.4	99.8	97.6	Aortic aneurysm, familial thoracic 10, 617168
LPL	147.2	100	100	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0
LRIT3	142.4	94.4	94.1	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP1	196	99.7	99.1	?Keratosis pilaris atrophicans, 604093
LRP2	176.3	100	99.8	Donnai-Barrow syndrome, 222448

LRP6	169.3	100	99.7	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRRC10	194	100	100	No OMIM phenotype Cardiomyopathy,dilated (Qu (2015) Mol Med Rep 12,3718)
LZTR1	134	100	99.4	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MAP2K1	92.3	99.8	95.6	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	107.9	97.6	89.2	Cardiofaciocutaneous syndrome 4, 615280
MAT2A	115.4	99.7	96.9	No OMIM phenotype Thoracic aortic aneurysms (Guo (2015) Am J Hum Genet 96, 170)
MED13L	134.6	100	99.6	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MEF2C	137.7	97.9	93.5	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFAP5	126.8	100	99.5	Aortic aneurysm, familial thoracic 9, 616166
MIB1	141.7	100	99.6	Left ventricular noncompaction 7, 615092
MLYCD	75.8	91.2	86.8	Malonyl-CoA decarboxylase deficiency, 248360
MMP2	164.4	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP21	93.3	90.2	84.6	Heterotaxy, visceral, 7, autosomal, 616749
MYBPC3	142.5	98.5	95.7	cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYBPHL	99.9	99.2	94.3	No OMIM phenotype
MYH11	132.6	100	99.3	Aortic aneurysm, familial thoracic 4, 132900
MYH6	113.3	99	96.1	Atrial septal defect 3, 614089 cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	111.4	99.4	96.8	cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160

				Scapuloperoneal syndrome, myopathic type, 181430
MYH7B	113.2	97.6	94.5	No OMIM phenotype ?Cardiomyopathy,left ventricular noncompaction (Esposito (2013) Orphanet J Rare Dis 8) ?Hearing loss (Haraksingh (2014) BMC Genomics 15,1155)
MYL2	134.6	98.7	90.1	cardiomyopathy, hypertrophic, 10, 608758
MYL3	103.1	100	100	cardiomyopathy, hypertrophic, 8, 608751
MYL4	159	100	100	?Atrial fibrillation, familial, 18, 617280
MYL7	130.9	100	100	No OMIM phenotype
MYLK	148.4	99.9	99.3	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	120.1	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYO1C	111.5	99.3	98.2	No OMIM phenotype ?Sensorineural hearing loss,bilateral (Zadro (2009) Biochim Biophys Acta 1792,27)
MYO6	89.7	98.1	92.3	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic Cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYOCD	182	100	100	No OMIM phenotype
MYOM1	149.5	99.8	98.4	No OMIM phenotype
MYOM2	156.5	100	99.4	No OMIM phenotype ?Tetralogy of Fallot (Grunert (2014) Hum Mol Genet 23,3115)
MYOT	139.6	99.3	95.4	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ1	97	100	100	No OMIM phenotype
MYOZ2	145.9	100	100	cardiomyopathy, hypertrophic, 16, 613838
MYPN	142.4	99.3	98.4	cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYZAP	131.5	95.2	91.1	No OMIM phenotype
NCOA1	152.6	100	99.6	No OMIM phenotype
NEBL	102.2	96.7	92.9	No OMIM phenotype Cardiomyopathy,dilated (Purejav (2010) J Am Coll Cardiol 56,1493)
NEXN	79.8	94.2	79.9	cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876

NGF	257.6	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NKX2-5	83.2	100	99.5	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	104.4	100	99.7	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NNT	136.9	98.6	97.1	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NODAL	160.7	100	99.9	Heterotaxy, visceral, 5, 270100
NOS1AP	192.2	100	100	No OMIM phenotype Long QT syndrome (Shigemizu (2015) PLoS One 10,e0130329) ?Obsessive-compulsive disorder (Delorme (2010) BMC Med Genet 11,108) {Cardiac repolarisation, association with} (Arking (2006) Nat Genet 38,644)
NOS3	107.6	95.3	91	{Alzheimer disease, late-onset, susceptibility to}, 104300 {Coronary artery spasm 1, susceptibility to}, 0 {Hypertension, pregnancy-induced}, 189800 {Hypertension, susceptibility to}, 145500 {Ischemic stroke, susceptibility to}, 601367 {Placental abruption}, 0
NOTCH1	137.5	99.1	98	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	172.4	100	99.9	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP3	115.6	99.4	96.1	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPPA	115.7	100	100	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPPB	160.7	100	100	No OMIM phenotype ?Hypertension (Zeng (2013) J Hum Hypertens 27,271) {Diabetes type 2,reduced risk,association with} (Meirhaeghe (2007) Hum Mol Genet 16,1343)
NR2F2	246.1	98.7	94.3	Congenital heart defects, multiple types, 4, 615779
NRAS	188.4	100	100	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470

				Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NUP155	115.1	97.6	92.3	?Atrial fibrillation 15, 615770
NUP37	168.4	98.5	93.4	No OMIM phenotype Atrial fibrillation (Haskell (2017) Circ Cardiovasc Genet 10, e001443)
OBSCN	159.3	99.3	98.2	No OMIM phenotype Cardiomyopathy,dilated (Marston (2015) PLoS One 10,e138568) Glioblastoma (Balakrishnan (2007) Cancer Res 67,3545) ?Breast cancer (Aloraifi (2015) FEBS J epub,epub) ?Schizophrenia (Fromer (2014) Nature 506,179) ?Cardiomyopathy,hypertrophic (Arimura (2007) Biochem Biophys Res Commun 362,281) Glioblastoma (Balakrishnan (2007) Cancer Res 67,3545)
PCCA	103.1	96.4	89.3	Propionicacidemia, 606054
PCCB	129.7	98.7	96.5	Propionicacidemia, 606054
PDLIM3	148.7	100	100	No OMIM phenotype Cardiomyopathy,dilated (Arola (2007) Mol Genet Metab 90,435 ?Cardiomyopathy, hypertrophic (Bagnall (2010) Int J Cardiol 145,601)
PGM1	133.9	100	99.9	Congenital disorder of glycosylation, type It, 614921
PHKA1	106.7	98.9	95.3	Muscle glycogenosis, 300559
PHYH	74.6	97.5	90.8	Refsum disease, 266500
PITX2	147.8	99.7	97.5	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PKD1L1	123.8	100	99.6	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	99.6	94.6	87.7	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	139	99.2	96.2	No OMIM phenotype
PLEC	114.1	99.7	98.7	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950

				Muscular dystrophy, limb-girdle, type 2Q, 613723
PLEKHM2	112.9	100	99.7	No OMIM phenotype Cardiomyopathy, dilated with left ventricular noncompaction (Muhammad (2015) Hum Mol Genet 24, 7227)
PLN	209.7	100	100	cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	137.9	99.8	97.5	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	113.2	99.7	97.4	Neutral lipid storage disease with myopathy, 610717
POMT1	155.7	99.7	98.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	111.1	98.9	97.5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PPA2	80.5	94.6	82.4	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARGC1A	142.7	99.9	99.3	No OMIM phenotype {Diabetes, type 2, association with}(Ek (2001) Diabetologia 44,2220)
PPP3CB	83.9	95.9	89.4	No OMIM phenotype
PPP3R2	221.4	100	100	No OMIM phenotype
PRDM16	161.7	100	99.1	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRKAG2	125.6	98.1	91.6	cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKG1	123.4	98.7	95.4	Aortic aneurysm, familial thoracic 8, 615436
PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAF1	127.3	100	99.7	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553

RANGRF	114	99.9	98.4	No OMIM phenotype Brugada syndrome (Selga (2015) PLoS One 10,e0132888 Histiocytoid Cardiomyopathy (Cataldo (2014)
RBM20	180.9	99.2	96.6	cardiomyopathy, dilated, 1DD, 613172
RIT1	165.6	100	100	Noonan syndrome 8, 615355
RPSA	88.9	100	99.7	Asplenia, isolated congenital, 271400
RYR1	120.7	96.8	93.7	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
RYR2	142.2	99.7	98.4	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
SCN10A	165.3	100	99.5	Episodic pain syndrome, familial, 2, 615551
SCN1B	168.3	97.1	96.1	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN2B	185.8	100	100	Atrial fibrillation, familial, 14, 615378
SCN3B	147.3	100	100	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	77.5	100	97.9	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	169.4	99	99	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 {Sudden infant death syndrome, susceptibility to}, 272120
SELENON	111.7	85.2	83.3	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310

SGCA	147.7	100	99.7	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	154.2	96.6	94.2	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	94.7	100	99.4	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCG	138.6	100	100	Muscular dystrophy, limb-girdle, type 2C, 253700
SHOC2	140.4	100	99.4	Noonan-like syndrome with loose anagen hair, 607721
SHROOM3	137.3	99.9	98.9	No OMIM phenotype Heterotaxy (Tariq (2011) Genome Biol 12,R91) ?Neural tube defects (Lemay (2015) J Med Genet 52,493) {Leukaemia risk,association with} (Rudd (2006) Blood 108,638)
SKI	85.3	96.4	90.8	Shprintzen-Goldberg syndrome, 182212
SLC22A5	153.3	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A20	110.3	100	99.7	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	134.2	100	100	Mitochondrial DNA depletion syndrome 12A (Cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC2A10	166.4	97.7	97.6	Arterial tortuosity syndrome, 208050
SLC8A1	198.2	99.7	99	No OMIM phenotype {Colorectal cancer,increased risk,association with} (Peters (2012) Hum Genet 131,217) ?Schizophrenia (Purcell (2014) Nature 506,185)
SLMAP	121.2	93.5	85.2	No OMIM phenotype Brugada syndrome (Ishikawa (2012) Circ Arrhythm Electrophysiol epub)
SMAD1	184	99.9	99.7	No OMIM phenotype
SMAD2	151.7	99.9	99.1	No OMIM phenotype Congenital heart disease (Zaidi (2013) Nature 498,220) Arterial aneurysms and dissections (Micha (2015) Hum Mutat 36,1145) Holoprosencephaly (Roessler (2008) Am J Hum Genet 83,18)
SMAD3	131.7	99.9	99.2	Loeys-Dietz syndrome 3, 613795
SMAD4	125.5	100	100	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	100.5	80	72	Aortic valve disease 2, 614823

				{Craniosynostosis 7, susceptibility to}, 617439
SMAD9	132.8	100	100	Pulmonary hypertension, primary, 2, 615342
SMYD1	140	100	100	No OMIM phenotype
SNTA1	97.2	82.4	77.3	Long QT syndrome 12, 612955
SOS1	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SRI	114.5	97.9	88.8	No OMIM phenotype Cardiomyopathy, hypertropic (Valvidia (2004) J Muscle Res Cell Motil 25,605)
SYNE1	136.6	98.2	97.6	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	123.1	98.6	96	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNPO2	195.8	99.7	99.4	No OMIM phenotype
TAB2	210.5	99.7	97.6	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	94	99.9	98.8	Barth syndrome, 302060
TBX1	75.3	77.1	67.4	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX20	142.8	99.9	99.3	Atrial septal defect 4, 611363
TBX5	141.3	100	100	Holt-Oram syndrome, 142900
TCAP	89	100	99.2	cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TDGF1	151.4	99.8	96.4	Forebrain defects, 0
TECRL	59.2	89.8	77.1	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	153.5	98.8	96.3	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TGFB2	176.9	100	99.9	Loeys-Dietz syndrome 4, 614816
TGFB3	171.5	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	173.4	93.7	93.6	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	193.5	100	99.9	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168

TLL1	140.1	100	99.9	Atrial septal defect 6, 613087
TMEM43	124.9	100	99.5	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMOD1	109.8	100	100	No OMIM phenotype
TMPO	117.8	98.6	94.4	?Cardiomyopathy, dilated, 1T, 613740
TNNC1	174.5	100	100	cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	86.7	98.1	86.5	?cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690
TNNI3K	118.8	98.8	96	?Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	106.3	100	99.9	cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TOR1AIP1	143.8	97.6	95.9	?Muscular dystrophy, limb-girdle, type 2Y, 617072
TPM1	132.9	99.7	97.9	cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TRDMT1	100	91.8	85.1	No OMIM phenotype
TRDN	71.8	83.5	70.7	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	118.5	100	99.6	No OMIM phenotype Hypertrophic Cardiomyopathy (Chen (2012) Circ Res 111,907)
TRPM4	109.4	99.7	98.5	Progressive familial heart block, type IB, 604559
TTN	187.8	98.2	97.2	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334
TTR	152.3	94.6	94.6	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680

TXNRD2	119.3	93.3	91.2	?Glucocorticoid deficiency 5, 617825
VCL	115.8	100	99.8	cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	138.5	100	99.7	No OMIM phenotype ?Schizphrenia (Fromer (2014) Nature 506,179)
XK	96.8	99.9	99.1	McLeod syndrome with or without chronic granulomatous disease, 300842
ZBTB17	142.2	100	100	No OMIM phenotype
ZFHX3	129.2	100	99.6	{Prostate cancer, susceptibility to, somatic}, 176807
ZFPM2	196.3	100	99.6	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	113.7	100	99.8	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 11th, 2018.
This list is accurate for panel version DG 2.14
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors