HEART GENE PANEL DG 2.9

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	141.5	99%	99%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCC6	121.4	93%	92%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	175	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAN	136.5	92%	85%	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACE	133.8	93%	92%	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase] {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to} {SARS, progression of} {Stroke, hemorrhagic}, 614519
ACSF3	146.1	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ACTA2	140.7	100%	99%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTC1	161.2	100%	99%	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424

ACTN1	164.8	100%	99%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	179.1	100%	100%	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACVR1	184.8	100%	99%	Fibrodysplasia ossificans progressiva, 135100
ACVR2B	171.9	99%	95%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS6	214.9	99%	99%	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179)
ADAMTS9	165.8	98%	96%	No OMIM phenotype
ADRB1	166.4	96%	89%	[Resting heart rate], 607276 {Congestive heart failure and beta-blocker response, modifier of}
ADRB2	147.4	100%	100%	Beta-2-adrenoreceptor agonist, reduced response to {Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665
AGL	184.8	99%	99%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGT	214.3	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}
AGTR1	187.9	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AKAP9	116.9	99%	96%	?Long QT syndrome-11, 611820

ALDH1A2	131.5	100%	99%	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	208.7	99%	99%	Alstrom syndrome, 203800
ANK2	168.6	100%	100%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKRD1	119.4	99%	96%	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)
ANKS6	95.2	93%	90%	Nephronophthisis 16, 615382
AP1B1	186.6	99%	98%	No OMIM phenotype
AP2B1	132.6	99%	98%	No OMIM phenotype
APBB1	152.8	100%	99%	No OMIM phenotype {Dementia alzheimer type,lower risk,association} (Hu (1998) Hum Genet 103,295)
ARMC4	135.4	91%	90%	Ciliary dyskinesia, primary, 23, 615451
ATP1A4	187.8	100%	99%	No OMIM phenotype
BAG3	140.5	100%	99%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954

BICC1	175.4	99%	99%	{Renal dysplasia, cystic, susceptibility to}, 601331
BMPR2	220.4	100%	99%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BRAF	86.4	91%	82%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706
C1orf127	115.5	99%	97%	No OMIM phenotype ?Autism (Lim (2013) Neuron 77,235)
C5orf42	154.1	99%	97%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CACNA1B	154.8	95%	92%	?Dystonia 23, 614860
CACNA1C	182.7	99%	99%	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	178.3	100%	99%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	111.5	96%	90%	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)
CACNB2	165.2	99%	98%	Brugada syndrome 4, 611876

CALM1	154.6	100%	100%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	65.8	68%	66%	Long QT syndrome 15, 616249
CALM3	118.8	100%	99%	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)
CALR3	165.4	100%	99%	?Cardiomyopathy, hypertrophic, 19, 613875
CAPN3	126.3	99%	98%	Muscular dystrophy, limb-girdle, type 2A, 253600
CASQ2	168.6	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV3	294	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
CBL	146	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CBS	134.1	98%	94%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200

CC2D2A	144.7	99%	97%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC151	134	100%	99%	Ciliary dyskinesia, primary, 30, 616037
CCDC39	112.3	99%	96%	Ciliary dyskinesia, primary, 14, 613807
CDKN1C	37.4	77%	63%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CEP290	92.8	95%	87%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CFC1	78.2	81%	69%	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	168.4	100%	99%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CITED2	122.5	100%	99%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CNTF	115.1	100%	100%	No OMIM phenotype {Ciliary neurotrophic factor deficiency} (Takahashi (1994) Nat Genet 7,79)
CNTRL	126.5	99%	96%	No OMIM phenotype Testicular cancer (Litchfield (2016) Nat Commun 7,13840)
COL3A1	124.9	98%	93%	Ehlers-Danlos syndrome, type IV, 130050

COL4A1	103.5	98%	94%	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL5A1	131.5	98%	95%	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	106.4	99%	97%	Ehlers-Danlos syndrome, classic type, 130000
CRELD1	113.4	99%	95%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRKL	175.7	100%	99%	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	120.6	99%	96%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CSRP3	113.2	100%	99%	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CTBP2	111.4	99%	96%	No OMIM phenotype ?Congenital heart disease (Glessner (2014) Circ Res 115,884)
CTF1	31.7	36%	21%	No OMIM phenotype Cardiomyopathy,dilated (Erdmann (2000) Hum Mutat 16,448)

CTLA4	212.7	100%	100%	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700
CTNNA3	167.9	100%	99%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CXADR	95.2	95%	89%	No OMIM phenotype
CXCR4	182.7	100%	99%	Myelokathexis, isolated WHIM syndrome, 193670
CYP11B2	190.1	100%	99%	Aldosterone to renin ratio raised Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}
DAW1	204.7	99%	97%	No OMIM phenotype
DCTN5	115.5	99%	95%	No OMIM phenotype ?Bipolar disorder (Rao (2016) Mol Psychiatry epub,epub)
DDX39B	20.9	72%	41%	No OMIM phenotype {Leprosy,susceptibility to,association with} (Ali (2012) Hum Genet 131,703)
DES	145.8	99%	99%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DMD	140.4	99%	98%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200

DNAAF3	95.4	98%	94%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	163.9	99%	99%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	151.6	99%	99%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	133.4	99%	98%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAJC19	115.5	98%	94%	3-methylglutaconic aciduria, type V, 610198
DNM2	145	99%	95%	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150
DOLK	189.5	100%	99%	Congenital disorder of glycosylation, type Im, 610768
DPP6	157.8	98%	95%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956

DRC1	108	99%	98%	Ciliary dyskinesia, primary, 21, 615294
DSC2	158.7	99%	97%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	149	99%	99%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSP	161.7	100%	99%	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	173.4	100%	100%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DYNC2H1	110.6	98%	91%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYX1C1	101.6	99%	96%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EDN1	166.6	100%	100%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}
EDNRA	229.1	100%	100%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300

EDNRB	151.4	98%	93%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EFEMP2	146.1	100%	99%	Cutis laxa, autosomal recessive, type IB, 614437
ELN	103.3	99%	98%	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
EMD	117.4	98%	94%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ETS1	138.7	99%	98%	No OMIM phenotype Congenital heart disease (Glessner (2014) Circ Res 115,884)
EYA4	189.6	100%	99%	Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FBN1	177.6	99%	99%	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection 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
FBN2	191.3	100%	99%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FHL1	98.4	98%	93%	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Hemophagocytic lymphohistiocytosis, familial, 1 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	176	99%	98%	No OMIM phenotype Cardiomyopathy,hypertrophic (Friedrich (2014) Basic Res Cardiol 109,451) ?Distal myopathy (Evila (2016) Neuromuscul Disord 26,7)
FKTN	155.6	99%	95%	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	160.7	100%	99%	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNC	188.5	100%	99%	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FOXC2	64.1	98%	88%	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 Lymphedema-distichiasis syndrome, 153400
FOXH1	59.6	99%	92%	No OMIM phenotype Congenital heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J Cardiol 145,83)
FOXJ1	81.8	99%	97%	No OMIM phenotype
FOXL1	93.3	87%	80%	No OMIM phenotype ?Hypoplastic left heart syndrome (Iascone (2012) Clin Genet 81,542)

FREM2	194.3	99%	99%	Fraser syndrome, 219000
FUZ	125	100%	99%	Neural tube defects, 182940
FXN	84.8	86%	76%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GAA	136.8	100%	99%	Glycogen storage disease II, 232300
GATA4	98.9	74%	63%	Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429 ?Testicular anomalies with or without congenital heart disease, 615542
GATA5	60.2	99%	95%	No OMIM phenotype Bicuspid aortic valve (Shi (2014) Int J Mol Med 33,1219) Atrial septal defect (Jiang (2013) Int J Cardiol 165,570) Atrial fibrillation (Gu (2012) Clinics (Sao Paulo) 67,1393) Atrioventricular septal defect, Down-syndrome-related (Ackerman (2012) Am J Hum Genet 91,646) Tetralogy of Fallot (Wei (2013) Int J Med Sci 10,34) Cardiomyopathy,dilated (Zhang (2015) Int J Mol Med 35,763)
GATA6	61.9	86%	73%	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	149.5	99%	95%	?Cardiomyopathy, dilated, 2B, 614672
GDF1	26	77%	56%	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854

GJA1	238.2	100%	100%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva, 133200 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	319.7	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJC1	210.1	100%	100%	No OMIM phenotype
GLA	83.5	99%	97%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GPD1L	184.1	100%	99%	Brugada syndrome 2, 611777
GTPBP3	139.5	100%	99%	Combined oxidative phosphorylation deficiency 23, 616198
H19	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
HAND1	99.3	100%	99%	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	54.3	96%	88%	No OMIM phenotype Tetralogy of Fallot (Topf (2014) PLoS One 9,e95453) Ventricular septal defect (Sun (2016) G3 (Bethesda) epub,epub) ?Congenital heart disease (Shen (2010) Chin Med J (Engl) 123,1623)

HCN1	155.1	100%	99%	Epileptic encephalopathy, early infantile, 24, 615871
HCN4	94.6	99%	96%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HECTD1	193	99%	98%	No OMIM phenotype ?Autism spectrum disorder (Wang (2016) Nat Commun 7,13316)
HEY2	172.1	99%	98%	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
HFE	148.7	100%	99%	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	135.7	100%	100%	Hemochromatosis type 2A,602390
HOOK1	98	97%	91%	No OMIM phenotype
HRAS	204.3	100%	99%	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
IDUA	120.5	92%	86%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016

IFNG	156	100%	99%	{AIDS, rapid progression to}, 609423 {Aplastic anemia}, 609135 {Hepatitis C virus, response to therapy of}, 609532 {TSC2 angiomyolipomas, renal, modifier of}, 613254 {Tuberculosis, protection against}, 607948
IFT140	124.5	99%	98%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT74	100	99%	93%	?Bardet-Biedl syndrome 20, 617119
IL10	152.8	100%	100%	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300
ILK	156.2	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)
IRX4	102.4	98%	93%	No OMIM phenotype Congenital heart defect (Cheng (2014) BMC Genomics 15,1127) {Prostate cancer,susceptibility to} (Nguyen (2012) Hum Mol Genet 21,2076)
JAG1	160.7	99%	98%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JPH2	106.7	97%	88%	Cardiomyopathy, hypertrophic, 17, 613873
JUP	160.5	100%	99%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KCNA5	160.1	100%	99%	Atrial fibrillation, familial, 7, 612240

KCND2	214.1	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	210.5	100%	98%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	538.9	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE1L	102.2	99%	93%	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)
KCNE2	165.4	100%	99%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	197	100%	100%	Brugada syndrome 6, 613119
KCNE4	114.6	80%	79%	No OMIM phenotype ?Periodic paralysis (Silva (2004) Arq Bras Endocrinol Metabol 48,196) {Atrial fibrillation, association with} (Zeng (2007) Cardiology 108,97)
KCNH2	118.6	95%	89%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ11	281.8	100%	100%	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ12	608.2	100%	99%	No OMIM phenotype

KCNJ2	210.8	100%	100%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ3	190.2	100%	100%	No OMIM phenotype {Schizophrenia, association with} (Yamada (2012) Hum Genet 131,443)
KCNJ5	219.7	100%	99%	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ8	180.4	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)
KCNMB1	134.6	100%	100%	{Hypertension, diastolic, resistance to}, 608622
KCNQ1	127.6	92%	89%	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	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	114.3	98%	95%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KIF7	95.3	95%	89%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KMT2D	158.6	100%	99%	Kabuki syndrome 1, 147920

KRAS	89.6	99%	99%	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
LAMA4	159.3	100%	99%	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	120.4	93%	91%	Danon disease, 300257
LDB3	146.5	95%	93%	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LEFTY2	46.5	89%	76%	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LIMS1	53.7	41%	34%	No OMIM phenotype

LMNA	95.6	97%	90%	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, 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210
LOX	148.1	99%	98%	No OMIM phenotype {Breast cancer,increased risk,in African American women,association with} (Min (2009) Cancer Res 69,6685) {Osteosarcoma, association with} (Liu (2012) PLoS One 7,e41610)
LRP1	219.2	99%	99%	?Keratosis pilaris atrophicans, 604093
LRP2	205.6	100%	99%	Donnai-Barrow syndrome, 222448
LRP6	178.9	100%	99%	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRRC10	207.4	100%	100%	No OMIM phenotype Cardiomyopathy,dilated (Qu (2015) Mol Med Rep 12,3718)
LTBP1	161.7	96%	94%	No OMIM phenotype ?Autism (Sanders (2012) Nature 485,237)
MAP2K1	106.2	99%	97%	Cardiofaciocutaneous syndrome 3, 615279

MAP2K2	122.2	99%	95%	Cardiofaciocutaneous syndrome 4, 615280
MCTP2	150.9	99%	98%	No OMIM phenotype Coarctation of the aorta (Lalani (2013) Hum Mol Genet 22,4339) ?Bicuspid aortic valve (Bonachea (2014) BMC Med Genomics 7,56)
MED13L	141.7	99%	99%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MEF2C	150.6	99%	96%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEGF8	142.7	99%	98%	Carpenter syndrome 2, 614976
MIB1	160.1	100%	99%	Left ventricular noncompaction 7, 615092
MICA	29.3	60%	47%	No OMIM phenotype Lung cancer (Huang (2011) Tissue antigens 78,297) {Ankylosing spondylitis, early onset, association with} (Amroun (2005) Hum Immunol 66,1057) {Rheumatoid arthritis, association with} (Kirsten (2009) Arthritis Res Ther 11,R60) {Cervical cancer,increased risk} (Chen (2013) J Natl Cancer Inst 105,624) {Oral squamous cell carcinoma, association with} (Tamaki (2007) J Oral Pathol Med 36,351) {Behcet disease,association with} (Mizuki (1997) Proc Natl Acad Sci USA 94,1298)
MICB	18.7	59%	34%	No OMIM phenotype {autism, association with} (Lim (2013) Neuron 77,235)
MMP21	115.9	93%	88%	Heterotaxy, visceral, 7, autosomal, 616749

MRPL3	74.4	92%	82%	Combined oxidative phosphorylation deficiency 9, 614582
MTO1	185.3	90%	88%	Combined oxidative phosphorylation deficiency 10, 614702
MYBPC3	161.5	99%	97%	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYH10	162.5	99%	99%	No OMIM phenotype Intrauterine growth restriction,microcephaly,developmental delay and hip dysplasia (Tuzovic (2013) Rare Dis 1,e26144) ?Intellectual disability (Hamdan (2014) PLoS Genet 10,e1004772) ?Autism spectrum disorder (Li (2016) Mol Psychiatry 21, 290)
MYH11	152.8	100%	99%	Aortic aneurysm, familial thoracic 4, 132900
MYH6	127.6	99%	97%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	123.2	99%	96%	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Liang distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430

MYH7B	129.6	99%	96%	No OMIM phenotype ?Cardiomyopathy,left ventricular noncompaction (Esposito (2013) Orphanet J Rare Dis 8) ?Hearing loss (Haraksingh (2014) BMC Genomics 15,1155)
MYL2	153.4	99%	96%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	124.4	100%	100%	Cardiomyopathy, hypertrophic, 8, 608751
MYL7	164.8	100%	100%	No OMIM phenotype
MYLK	168.7	99%	99%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	121.5	100%	99%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYO1C	122	99%	98%	No OMIM phenotype ?Sensorineural hearing loss,bilateral (Zadro (2009) Biochim Biophys Acta 1792,27)
MYOM2	182.6	100%	99%	No OMIM phenotype ?Tetralogy of Fallot (Grunert (2014) Hum Mol Genet 23,3115)
MYOT	172.4	99%	98%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ1	99.8	100%	99%	No OMIM phenotype
MYOZ2	194.6	100%	100%	Cardiomyopathy, hypertrophic, 16, 613838

MYPN	165.3	99%	98%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248
MYZAP	146.3	96%	93%	No OMIM phenotype
NAT8	183.8	100%	100%	No OMIM phenotype ?Microalbuminuria and dysplastic kidney (Carmichael (2013) Clin Genet 84,213) ?Altered N-acetylornithine metabolism (Yu (2014) PLoS Genet 10,e1004212
NDST1	231.1	100%	100%	Mental retardation, autosomal recessive 46, 616116
NEBL	128.3	98%	96%	No OMIM phenotype Cardiomyopathy,dilated (Purejav (2010) J Am Coll Cardiol 56,1493)
NEK8	192.1	100%	100%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NEXN	87.1	93%	83%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NFATC1	137.1	99%	97%	No OMIM phenotype Tricuspid atresia (Abdul-Sater(2012) PLoS One 7,e49532) Congenital heart disease (Glessner (2014) Circ Res 115,884) ?Biscuspid aortic valve (Bonachea (2014) BMC Med Genomics 7,56) ?Tetralogy of Fallot (Silversides (2012) PLoS Genet 8, e1002843) ?Ventricular septal defect (Zhao (2013) Am J Med Genet A 161,3087)
NFATC4	110.1	97%	95%	No OMIM phenotype {Cardiac hypertrophy,protection,association} (Poirier (2003) Eur J Hum Genet 11,659
NFKBIL1	10.4	44%	14%	{Rheumatoid arthritis, susceptibility to}, 180300

NGF	268.3	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NKX2-5	96.3	100%	99%	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 Tetrology of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	126.1	100%	98%	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NODAL	155.4	100%	100%	Heterotaxy, visceral, 5, 270100
NOS1AP	209.9	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	118.9	94%	91%	{Alzheimer disease, late-onset, susceptibility to}, 104300 {Coronary artery spasm 1, susceptibility to} {Hypertension, pregnancy-induced}, 189800 {Hypertension, susceptibility to}, 145500 {Ischemic stroke, susceptibility to}, 601367 {Placental abruption}
NOTCH1	154.9	99%	98%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	180.3	100%	99%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPA	124	100%	100%	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745

NPPB	180.8	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	255.8	99%	95%	Congenital heart defects, multiple types, 4, 615779
NRAS	203.3	100%	100%	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 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
NSD1	181.1	100%	100%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
OBSCN	172.1	99%	98%	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)
PAFAH1B1	125	92%	87%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PCSK5	180.2	100%	99%	No OMIM phenotype ?Low HDL cholesterol (Motazacker (2013) Arterioscler Thromb Vasc Biol 33,1521) ?VACTERL (Nakamura (2015) BMC Res Notes 8,228)
PDE2A	128.5	99%	98%	No OMIM phenotype

PDLIM3	193.6	100%	100%	No OMIM phenotype Cardiomyopathy,dilated (Arola (2007) Mol Genet Metab 90,435 ?Cardiomyopathy, hypertrophic (Bagnall (2010) Int J Cardiol 145,601)
PITX2	146.1	99%	98%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PKD1	30.1	41%	33%	Polycystic kidney disease, adult type I, 173900
PKD1L1	137.6	99%	99%	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	112.7	96%	91%	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	145.8	98%	95%	No OMIM phenotype
PLA2G7	158.1	100%	99%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLEC	122.1	99%	99%	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 ?Epidermolysis bullosa simplex with nail dystrophy, 616487
PLN	252.7	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874

PLXND1	135.4	97%	94%	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6) Truncus arteriosus (Ta-Shma (2013) Am J Med Genet A 161,3115) {Diabetic nephropathy,association with} (McKnight (2009) Hugo J 3,77)
PNN	141	99%	97%	No OMIM phenotype
PPARGC1A	172	99%	99%	No OMIM phenotype {Diabetes, type 2, association with}(Ek (2001) Diabetologia 44,2220)
PRDM1	186.4	100%	99%	No OMIM phenotype {Chrohn's disease,increased risk,association with} (Ellinghaus (2013) Gastroenterology 145,339 {Ulcerative colitis,reduced risk,association with} (Ellinghaus (2013) Gastroenterology 145,339 ?Colorectal cancer (Zhang (2015) World J Gastroenterol 21,4136) ?Truncus arteriosus (Shaheen (2015) J Med Genet 52,322)
PRDM16	180	99%	99%	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRICKLE1	138.6	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRKAG2	146.1	99%	95%	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKG1	133.1	99%	96%	Aortic aneurysm, familial thoracic 8, 615436
PSKH1	249.9	100%	100%	No OMIM phenotype
PTK7	168.8	100%	99%	No OMIM phenotype ?Autism (Sanders (2012) Nature 485,237) ?Neural tube defects (Wang (2015) Birth Defects Res A Clin Mol Teratol epub)

PTPLA	66.3	75%	67%	No OMIM phenotype ?Myopathy,congenital (Muhammad (2013) Hum Mol Genet 22,5229)
PTPN11	105.7	98%	93%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN22	162.4	99%	94%	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRC	127.6	96%	89%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitic C virus, susceptibility to}, 609532
PTPRM	166.8	100%	99%	No OMIM phenotype
RAF1	144.9	100%	99%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RANGRF	117	99%	97%	No OMIM phenotype Brugada syndrome (Selga (2015) PLoS One 10,e0132888 Histiocytoid cardiomyopathy (Cataldo (2014)
RBM20	194.8	99%	97%	Cardiomyopathy, dilated, 1DD, 613172
RIT1	190.3	100%	100%	Noonan syndrome 8, 615355
ROBO1	197.4	100%	99%	No OMIM phenotype Breast and colorectal cancer (Villacis (2015) Tumour Biol epub, epub) ?Developmental dyslexia (Hannula-Jouppi (2005) PLoS Genet 1,e50) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89, 476)
ROBO2	167.1	99%	97%	Vesicoureteral reflux 2, 610878

RPSA	89	100%	99%	Asplenia, isolated congenital, 271400
RYR2	167.3	99%	99%	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
SCN10A	187	99%	99%	Episodic pain syndrome, familial, 2, 615551
SCN1B	189.8	97%	96%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2B	211.9	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3B	163.5	100%	100%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	81.8	99%	93%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	181.2	100%	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
SCNN1B	172.3	100%	99%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350

SCNN1G	160.7	99%	97%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCO2	126.5	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SEMA3D	169	99%	98%	No OMIM phenotype Congenital heart defects (Sanchez-Castro (2015) Hum Mutat 36,30) Hirschsprung disease (Jiang (2015) Am J Hum Genet 96,581) ?Total anomalous pulmonary venous connection (Degenhardt (2013) Nat Med 19,760) ?Tetralogy of Fallot (Siversides (2012) PLoS Genet 8,e1002843)
SGCA	173.2	100%	99%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	192.5	97%	96%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	109.9	99%	98%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCE	110.4	95%	92%	Dystonia-11, myoclonic, 159900
SGCG	141.7	100%	99%	Muscular dystrophy, limb-girdle, type 2C, 253700
SHOC2	150.5	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM3	150.3	99%	98%	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	96.2	98%	96%	Shprintzen-Goldberg syndrome, 182212
SLC22A5	171.6	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC25A4	142.3	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	187.1	100%	99%	Arterial tortuosity syndrome, 208050
SLC8A1	226.4	99%	98%	No OMIM phenotype {Colorectal cancer,increased risk,association with} (Peters (2012) Hum Genet 131,217) ?Schizophrenia (Purcell (2014) Nature 506,185)
SLMAP	156.8	96%	88%	No OMIM phenotype Brugada syndrome (Ishikawa (2012) Circ Arrhythm Electrophysiol epub)
SMAD2	165.3	100%	99%	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	151.7	99%	99%	Loeys-Dietz syndrome 3, 613795
SMAD6	111	91%	83%	Aortic valve disease 2, 614823
SMARCA4	165.4	100%	99%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325

SMYD1	137.8	100%	99%	No OMIM phenotype
SNTA1	104.9	85%	77%	Long QT syndrome 12, 612955
SNTB1	124.9	100%	99%	No OMIM phenotype
SNX17	171.7	100%	99%	No OMIM phenotype
SOD2	239.6	100%	100%	{Microvascular complications of diabetes 6}, 612634
SOS1	120.1	98%	95%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SUFU	146	99%	97%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SYNE1	172	99%	99%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	135.1	99%	97%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNPO2	183.4	99%	98%	No OMIM phenotype
TAB1	166.5	100%	99%	No OMIM phenotype
TAB2	232.8	99%	98%	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	123.4	99%	98%	Barth syndrome, 302060

TBC1D32	100.8	98%	94%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TBX1	90.6	79%	70%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX20	137.1	100%	99%	Atrial septal defect 4, 611363
TBX3	95.1	99%	97%	Ulnar-mammary syndrome, 181450
TBX5	145.9	100%	100%	Holt-Oram syndrome, 142900
TCAP	99.8	99%	97%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TDGF1	170.5	99%	95%	Forebrain defects
TFAP2B	145.4	98%	96%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TGFB1	84.7	99%	98%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	203	100%	99%	Loeys-Dietz syndrome 4, 614816
TGFB3	177.7	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	204.7	94%	93%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	212.4	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168

TLL1	173.9	100%	100%	Atrial septal defect 6, 613087
TMEM43	145.6	99%	99%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM67	93.3	95%	89%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMOD1	127.7	100%	99%	No OMIM phenotype
TMPO	146.3	99%	96%	?Cardiomyopathy, dilated, 1T, 613740
TNF	20.5	81%	45%	{Asthma, susceptibility to}, 600807 {Dementia, vascular, susceptibility to} {Malaria, cerebral, susceptibility to}, 611162 {Migraine without aura, susceptibility to}, 157300 {Septic shock, susceptibility to}
TNNC1	208.9	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	122.5	99%	95%	Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880
TNNI3K	155.9	100%	99%	?Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	117.4	100%	99%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494

TPM1	155.6	99%	98%	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TRDN	85.6	90%	79%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	153.6	99%	99%	No OMIM phenotype Hypertrophic cardiomyopathy (Chen (2012) Circ Res 111,907)
TRPM4	128	99%	98%	Progressive familial heart block, type IB, 604559
TTN	236.7	98%	97%	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, type 2J, 608807 Myopathy, early-onset, with fatal cardiomyopathy, 611705 Myopathy, proximal, with early respiratory muscle involvement, 603689 Tibial muscular dystrophy, tardive, 600334
TTR	184.7	100%	100%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
VCL	117.4	100%	99%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	160.9	100%	100%	No OMIM phenotype ?Schizphrenia (Fromer (2014) Nature 506,179)
ZBTB14	222.8	100%	99%	No OMIM phenotype
ZBTB17	164.4	100%	100%	No OMIM phenotype

ZEB2	195.1	100%	99%	Mowat-Wilson syndrome, 235730
ZFPM2	208.3	100%	99%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	127.1	100%	99%	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 : April 14th 2017
This list is accurate for panel version DG 2.9

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