HEART GENE PANEL DG 2.12 (400 genes)

Gene	*Median*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
A2ML1	130.9	100	99	No OMIM phenotype Noonan-like syndrome (Vissers et al. 2015) Noonan syndrome (van Trier (2015) Int J Pediatr Otorhinolaryngol, epub) Otitis media, susceptibility to (Santos-Cortez (2015) Nat Genet 47,917)
AARS2	126.4	99	99	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCC6	116.6	93	92	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	157.8	99	99	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAD9	135.4	98	95	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACADVL	118.9	98	95	VLCAD deficiency, 201475
ACSF3	128.8	99	99	Combined malonic and methylmalonic aciduria, 614265
ACTA1	100	99	95	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 ?Myopathy, scapulohumeroperoneal, 616852
ACTA2	138	100	99	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTC1	164.4	100	99	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTN1	143.9	100	99	Bleeding disorder, platelet-type, 15, 615193

ACTN2	159.6	100	100	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACVR1	165.2	100	100	Fibrodysplasia ossificans progressiva, 135100
ACVR2B	140.6	97	94	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS9	141.5	97	96	No OMIM phenotype
ADCY5	129.5	92	89	Dyskinesia, familial, with facial myokymia, 606703
ADRB1	155.4	97	89	[Resting heart rate], 607276 {Congestive heart failure and beta-blocker response, modifier of}
AGK	112.2	99	96	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	145.9	99	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	109.6	99	95	Lipodystrophy, congenital generalized, type 1, 608594
AGRN	115.1	95	89	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	214.4	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}
AGTR1	149.9	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AKAP9	99	98	95	?Long QT syndrome-11, 611820
ALDH1A2	114.7	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	179.4	99	99	Alstrom syndrome, 203800
ALPK3	98.9	94	92	No OMIM phenotype
ANK2	160.4	100	99	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKRD1	101.8	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	91.8	92	88	Nephronophthisis 16, 615382

ANO5	142.1	99	96	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
APBB1	140	100	99	No OMIM phenotype {Dementia alzheimer type,lower risk,association} (Hu (1998) Hum Genet 103,295)
ATP1A4	161.6	100	99	No OMIM phenotype
ATPAF2	101.5	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	136.9	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BGN	129.9	100	99	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BOLA3	50.1	92	81	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAF	74.3	87	77	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
BRCC3	53.7	84	63	No OMIM phenotype
BSCL2	113.7	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
C5orf42	122.4	98	95	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CACNA1C	155.1	99	99	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	152.9	100	99	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	82.6	93	84	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	112.1	99	97	Retinal cone dystrophy 4, 610478
CACNB2	151	99	96	Brugada syndrome 4, 611876
CALM1	114.2	100	99	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	54.1	67	65	Long QT syndrome 15, 616249
CALM3	117.5	99	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	137.4	100	99	?Cardiomyopathy, hypertrophic, 19, 613875
CAPN3	109.9	98	96	Muscular dystrophy, limb-girdle, type 2A, 253600
CARD6	149.8	97	97	No OMIM phenotype
CASQ2	143.2	99	99	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV1	265.7	100	100	Pulmonary hypertension, primary, 3, 615343 ?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721
CAV3	305.2	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	133.2	99	98	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CBS	114.4	97	91	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D2A	127.4	99	97	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC11	146.5	97	94	Heterotaxy, visceral, 6, autosomal recessive, 614779
CDH2	137.5	98	97	No OMIM phenotype
CDKN1C	21.3	68	51	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CFC1	76.7	83	73	Heterotaxy, visceral, 2, autosomal, 605376

CHD7	150.9	99	98	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHKB	98.7	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	142.6	100	99	No OMIM phenotype
CITED2	114.1	100	99	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	104.2	97	92	Ehlers-Danlos syndrome, type IV, 130050
COL4A1	92.8	97	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	115.4	97	95	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	89	99	97	Ehlers-Danlos syndrome, classic type, 130000
COQ2	89.2	96	93	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COX15	98.7	100	99	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX6B1	159.9	100	100	Mitochondrial complex IV deficiency, 220110
CPT1A	169.5	100	98	CPT deficiency, hepatic, type IA, 255120
CPT2	166.7	98	97	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	114.6	99	97	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRKL	166.9	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	125.9	99	98	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869

CSRP3	103	100	99	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CTBP2	105.2	99	96	No OMIM phenotype ?Congenital heart disease (Glessner (2014) Circ Res 115,884)
CTF1	24.5	28	20	No OMIM phenotype Cardiomyopathy,dilated (Erdmann (2000) Hum Mutat 16,448)
CTNNA3	138.2	100	99	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CXADR	95.1	95	88	No OMIM phenotype
DAG1	221.1	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DAW1	158.7	98	94	No OMIM phenotype
DCTN5	107.9	99	92	No OMIM phenotype ?Bipolar disorder (Rao (2016) Mol Psychiatry epub,epub)
DES	121	99	98	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DMD	113.7	99	97	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMPK	117.8	99	97	Myotonic dystrophy 1, 160900
DNAH5	123.8	99	98	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAJC19	97.8	98	89	3-methylglutaconic aciduria, type V, 610198
DNM2	127.9	97	94	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	203	100	99	Congenital disorder of glycosylation, type Im, 610768
DPM3	184.3	100	100	Congenital disorder of glycosylation, type Io, 612937
DPP6	145.7	96	94	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	128.3	99	96	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476

DSG2	140.7	99	98	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSP	154.1	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	156.1	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DYNC2H1	90.4	96	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYSF	133.2	100	99	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, type 2B, 253601 Myopathy, distal, with anterior tibial onset, 606768
DYX1C1	79.6	96	84	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EDN1	145.4	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}
EDNRA	218.7	100	99	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EEF1A2	178	98	93	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP2	121.1	100	99	Cutis laxa, autosomal recessive, type IB, 614437
ELN	91.2	99	97	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
EMD	101.1	99	97	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENG	128.9	97	93	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	134.9	92	83	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	126.1	99	97	Vici syndrome, 242840
ETS1	112.7	99	98	No OMIM phenotype Congenital heart disease (Glessner (2014) Circ Res 115,884)

EYA4	160.5	100	99	Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FAH	151.6	100	99	Tyrosinemia, type I, 276700
FBN1	159.8	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	161.8	100	99	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXO32	150.8	100	100	No OMIM phenotype
FGF12	95.6	99	96	?Epileptic encephalopathy, early infantile, 47, 617166
FGF13	104.2	99	97	No OMIM phenotype
FHL1	87.8	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	149.8	99	98	No OMIM phenotype Cardiomyopathy,hypertrophic (Friedrich (2014) Basic Res Cardiol 109,451) ?Distal myopathy (Evila (2016) Neuromuscul Disord 26,7)
FHOD3	135.3	99	98	No OMIM phenotype
FKRP	94.6	100	99	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	119.9	99	94	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	139.2	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	165.3	100	99	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLT4	156.1	98	97	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100
FOXC2	44.3	95	79	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 Lymphedema-distichiasis syndrome, 153400
FOXD4	3.1	25	13	No OMIM phenotype
FOXE3	20.8	69	48	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256 Cataract 34,multiple types, 612968
FOXH1	47.4	98	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)
FOXL1	80.3	83	76	No OMIM phenotype ?Hypoplastic left heart syndrome (Iascone (2012) Clin Genet 81,542)
FREM2	182.5	100	99	Fraser syndrome, 219000
FXN	75.3	85	75	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GAA	128.7	100	99	Glycogen storage disease II, 232300

GATA4	87.5	68	60	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	44.4	98	84	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	61.6	83	71	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	145.4	99	97	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDF1	19.4	64	48	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854
GDF2	163.3	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GJA1	246.5	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	268.2	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770

GJC1	192.3	100	100	No OMIM phenotype
GLA	82	99	97	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	91.6	99	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GMPPB	229.1	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	126	99	98	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	153.9	100	99	Nonaka myopathy, 605820 Sialuria, 269921
GNPTAB	167.6	98	97	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GPD1L	138.4	100	98	Brugada syndrome 2, 611777
H19	NC	NC	NC	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
HADHA	84.1	96	90	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	80.4	92	79	Trifunctional protein deficiency, 609015
HAND1	85	100	98	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	32.3	87	68	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	123.5	99	97	Epileptic encephalopathy, early infantile, 24, 615871

HCN4	79.6	98	91	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HECTD1	172.9	98	96	No OMIM phenotype ?Autism spectrum disorder (Wang (2016) Nat Commun 7,13316)
HEY2	146.2	99	92	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
HFE	142	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	117	100	100	Hemochromatosis type 2A,602390
HOOK1	76.5	95	86	No OMIM phenotype
HRAS	164.9	99	98	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	63.2	89	77	No OMIM phenotype
IDUA	121.3	89	82	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFNG	130.5	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
IFT74	80.2	97	88	?Bardet-Biedl syndrome 20, 617119
IL10	125.4	100	99	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300

ILK	159	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	73.9	83	66	No OMIM phenotype
IRX4	91.5	95	91	No OMIM phenotype Congenital heart defect (Cheng (2014) BMC Genomics 15,1127) {Prostate cancer,susceptibility to} (Nguyen (2012) Hum Mol Genet 21,2076)
ISPD	104.2	95	84	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITGB1BP2	75.6	99	95	No OMIM phenotype
ITPA	120.4	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAG1	148.6	98	97	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JPH2	87.6	90	75	Cardiomyopathy, hypertrophic, 17, 613873
JUP	145.3	100	99	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KCNA5	143.7	99	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	182.9	99	99	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	463.7	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE1L	88.7	97	90	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	181.5	100	100	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	177.2	100	100	Brugada syndrome 6, 613119

KCNE4	84.9	79	77	No OMIM phenotype ?Periodic paralysis (Silva (2004) Arq Bras Endocrinol Metabol 48,196) {Atrial fibrillation, association with} (Zeng (2007) Cardiology 108,97)
KCNH2	102.8	92	84	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ11	300.2	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	575.3	100	100	No OMIM phenotype
KCNJ2	229.3	100	100	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ3	167.6	100	100	No OMIM phenotype {Schizophrenia, association with} (Yamada (2012) Hum Genet 131,443)
KCNJ5	194.5	100	99	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	165.6	98	96	Pulmonary hypertension, primary, 4, 615344
KCNMB1	116.4	100	100	{Hypertension, diastolic, resistance to}, 608622
KCNQ1	114.9	93	90	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

KIF7	85.8	93	88	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KLF10	141.7	100	99	No OMIM phenotype
KLHL24	193.1	100	100	Epidermolysis bullosa simplex, generalized, with scarring and hair loss ,617294
KMT2D	142.2	99	99	Kabuki syndrome 1, 147920
KRAS	64.6	99	98	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	143.6	99	99	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMA4	132.6	100	99	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	106.8	92	91	Danon disease, 300257
LARGE	142.6	100	99	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LDB3	127.4	95	93	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	77	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LIMS1	51.2	36	29	No OMIM phenotype
LMF1	132.8	99	97	Lipase deficiency, combined, 246650

LMNA	89.4	98	91	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
LMOD1	177.6	100	100	No OMIM phenotype
LOX	104.6	99	97	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)
LPL	147.1	100	100	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11]
LRIT3	142.6	94	94	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP1	196.4	99	99	?Keratosis pilaris atrophicans, 604093
LRP2	176.4	100	99	Donnai-Barrow syndrome, 222448
LRP6	169.5	100	99	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRRC10	193.4	100	100	No OMIM phenotype Cardiomyopathy,dilated (Qu (2015) Mol Med Rep 12,3718)
LTBP1	138.2	95	93	No OMIM phenotype ?Autism (Sanders (2012) Nature 485,237)
LTBP2	104.7	99	97	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 Weill-Marchesani syndrome 3, recessive, 614819
LZTR1	134.3	100	99	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670

MAP2K1	92.3	99	95	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	108	97	89	Cardiofaciocutaneous syndrome 4, 615280
MAT2A	115.3	99	96	No OMIM phenotype
MED12	106.5	98	94	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13L	134.7	100	99	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MEF2C	137	97	93	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEGF8	127.9	99	98	Carpenter syndrome 2, 614976
MFAP5	126.7	100	99	Aortic aneurysm, familial thoracic 9, 616166
MIB1	141.7	100	99	Left ventricular noncompaction 7, 615092
MLYCD	75.8	91	86	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	93.4	90	84	Heterotaxy, visceral, 7, autosomal, 616749
MURC	159.4	100	100	No OMIM phenotype
MYBPC3	142.8	98	95	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYBPHL	100	99	94	No OMIM phenotype
MYH10	131.6	99	98	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	132.8	100	99	Aortic aneurysm, familial thoracic 4, 132900
MYH6	113.5	99	96	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090

MYH7	111.5	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	113.4	97	94	No OMIM phenotype ?Cardiomyopathy,left ventricular noncompaction (Esposito (2013) Orphanet J Rare Dis 8) ?Hearing loss (Haraksingh (2014) BMC Genomics 15,1155)
MYL2	134.9	98	90	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	103.2	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYL7	131.1	100	100	No OMIM phenotype
MYLK	148.6	99	99	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	120.3	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYO6	89.7	98	92	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYOCD	182.1	100	100	No OMIM phenotype
MYOM1	149.7	99	98	No OMIM phenotype
MYOM2	156.6	100	99	No OMIM phenotype ?Tetralogy of Fallot (Grunert (2014) Hum Mol Genet 23,3115)
MYOT	139.4	99	95	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ1	97.3	100	100	No OMIM phenotype
MYOZ2	146	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	142.6	99	98	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248
MYZAP	131.5	95	91	No OMIM phenotype
NCOA1	152.5	100	99	No OMIM phenotype
NDST1	201.7	100	100	Mental retardation, autosomal recessive 46, 616116
NEBL	103.7	96	92	No OMIM phenotype

				Cardiomyopathy,dilated (Purejav (2010) J Am Coll Cardiol 56,1493)
NEK8	171.6	100	99	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NEXN	78	92	78	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	129.8	93	90	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFATC1	136.7	99	96	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	102.3	96	95	No OMIM phenotype {Cardiac hypertrophy,protection,association} (Poirier (2003) Eur J Hum Genet 11,659
NGF	258.2	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NKX2-5	83.2	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	104	100	99	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NODAL	160.9	100	99	Heterotaxy, visceral, 5, 270100
NOS1AP	192.5	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	107.8	95	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	137.8	99	98	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	172.7	100	99	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP3	114.3	99	95	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPPA	114.8	100	100	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPPB	160.8	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	246.2	98	94	Congenital heart defects, multiple types, 4, 615779
NRAS	188.2	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	155.4	100	100	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550

OBSCN	159.6	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	105	89	81	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PCCA	103.1	96	89	Propionicacidemia, 606054
PCCB	129.9	98	96	Propionicacidemia, 606054
PCSK5	165.7	100	100	No OMIM phenotype ?Low HDL cholesterol (Motazacker (2013) Arterioscler Thromb Vasc Biol 33,1521) ?VACTERL (Nakamura (2015) BMC Res Notes 8,228)
PDE2A	111.7	100	99	No OMIM phenotype
PDLIM3	148.6	100	100	No OMIM phenotype Cardiomyopathy,dilated (Arola (2007) Mol Genet Metab 90,435 ?Cardiomyopathy, hypertrophic (Bagnall (2010) Int J Cardiol 145,601)
PITX2	147.9	99	97	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PKD1L1	123.9	100	99	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	99.7	94	87	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	139.7	99	96	No OMIM phenotype
PLA2G7	124.4	99	97	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLEC	114.3	99	98	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
PLEKHM2	113	100	99	No OMIM phenotype Cardiomyopathy, dilated with left ventricular noncompaction (Muhammad (2015) Hum Mol Genet 24,

				7227)
PLN	209.3	100	100	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	134.3	99	97	Ehlers-Danlos syndrome, type VI, 225400
PLXND1	111	96	93	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	144.5	99	97	No OMIM phenotype
POMGNT1	126.8	99	97	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMT1	156	99	98	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
PPARGC1A	143.3	99	99	No OMIM phenotype {Diabetes, type 2, association with}(Ek (2001) Diabetologia 44,2220)
PRDM1	173.9	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	161.5	99	99	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRICKLE1	117.3	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRKAG2	125.8	98	91	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKG1	123.6	98	95	Aortic aneurysm, familial thoracic 8, 615436
PSKH1	251.6	100	100	No OMIM phenotype
PTPLA	57.1	72	60	No OMIM phenotype ?Myopathy,congenital (Muhammad (2013) Hum Mol Genet 22,5229)

PTPN11	103.4	97	92	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN22	134.4	98	91	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRC	101.6	93	86	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitic C virus, susceptibility to}, 609532
PTPRM	150.7	100	99	No OMIM phenotype
RAF1	127.4	100	99	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RANGRF	114.2	99	98	No OMIM phenotype Brugada syndrome (Selga (2015) PLoS One 10,e0132888 Histiocytoid cardiomyopathy (Cataldo (2014)
RASA2	86.1	92	80	No OMIM phenotype Noonan syndrome (Chen (2014) Proc Natl Acad Sci USA 111, 11473)
RBM20	181.2	99	96	Cardiomyopathy, dilated, 1DD, 613172
RIT1	165.5	100	100	Noonan syndrome 8, 615355
ROBO1	160.7	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	155.3	98	96	Vesicoureteral reflux 2, 610878
RPSA	88.6	100	99	Asplenia, isolated congenital, 271400
RRAS	116.5	89	81	No OMIM phenotype
RYR2	142.3	99	98	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
SCN10A	165.5	100	99	Episodic pain syndrome, familial, 2, 615551
SCN1B	168.8	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	186.2	100	100	Atrial fibrillation, familial, 14, 615378
SCN3B	147.5	100	100	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	77.7	100	97	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	171.4	100	100	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	149	100	99	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	139.6	99	97	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCO2	113.3	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SEMA3D	142.4	99	96	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)
SEPN1	111.9	85	83	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SGCA	144.8	100	99	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	154.1	96	94	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	94.8	100	99	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, type 2F, 601287
SGCE	88.9	93	90	Dystonia-11, myoclonic, 159900
SGCG	138.9	100	100	Muscular dystrophy, limb-girdle, type 2C, 253700

SHOC2	140.4	100	99	Noonan-like syndrome with loose anagen hair, 607721
SHROOM3	137.6	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	85.5	96	90	Shprintzen-Goldberg syndrome, 182212
SLC22A5	153.5	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A4	134	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	170.6	100	99	Arterial tortuosity syndrome, 208050
SLC8A1	197.3	99	99	No OMIM phenotype {Colorectal cancer,increased risk,association with} (Peters (2012) Hum Genet 131,217) ?Schizophrenia (Purcell (2014) Nature 506,185)
SLMAP	126.4	93	85	No OMIM phenotype Brugada syndrome (Ishikawa (2012) Circ Arrhythm Electrophysiol epub)
SMAD1	183.9	99	99	No OMIM phenotype
SMAD2	151.5	99	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	132	99	99	Loeys-Dietz syndrome 3, 613795
SMAD4	121.2	99	98	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	100.5	81	73	Aortic valve disease 2, 614823
SMAD9	132.7	100	100	Pulmonary hypertension,primary,615342 Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)
SMARCA4	144.8	100	99	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMYD1	140.2	100	100	No OMIM phenotype
SNTA1	97.1	82	77	Long QT syndrome 12, 612955
SNX17	159	100	100	No OMIM phenotype

SOD2	212.8	100	100	{Microvascular complications of diabetes 6}, 612634
SOS1	95.5	96	90	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SUFU	122.7	100	99	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SYNE1	139.1	99	99	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	122.9	98	96	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNPO2	196.7	100	99	No OMIM phenotype
TAB1	158.1	100	99	No OMIM phenotype
TAB2	209.6	99	97	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	94.9	99	98	Barth syndrome, 302060
TBC1D32	81.2	96	91	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TBX1	75.5	77	67	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX20	143.2	99	99	Atrial septal defect 4, 611363
TBX3	80.6	99	95	Ulnar-mammary syndrome, 181450
TBX5	141.6	100	100	Holt-Oram syndrome, 142900
TCAP	89.3	100	99	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, type 2G, 601954
TDGF1	151.5	99	96	Forebrain defects
TFAP2B	155.6	98	96	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TGFB1	87.1	99	95	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	176.9	100	99	Loeys-Dietz syndrome 4, 614816
TGFB3	171.8	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	173.5	93	93	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800

TGFBR2	193.8	100	99	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TLL1	140	100	99	Atrial septal defect 6, 613087
TMEM43	125	100	99	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM67	72.9	93	83	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMOD1	109.9	100	100	No OMIM phenotype
TMPO	117.7	98	94	?Cardiomyopathy, dilated, 1T, 613740
TNNC1	175.2	100	100	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	86.8	98	86	Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880
TNNI3K	118.3	98	95	?Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	105.8	100	99	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TPM1	137.4	99	97	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TRDN	68.2	83	69	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM32	141.3	100	100	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRIM63	119	100	99	No OMIM phenotype Hypertrophic cardiomyopathy (Chen (2012) Circ Res 111,907)
TRPM4	109.3	99	98	Progressive familial heart block, type IB, 604559
TSFM	123.2	100	100	Combined oxidative phosphorylation deficiency 3, 610505

TTN	187.7	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	159.7	100	100	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
VCL	115.8	100	99	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	138.4	100	99	No OMIM phenotype ?Schizphrenia (Fromer (2014) Nature 506,179)
ZBTB14	193.2	100	99	No OMIM phenotype
ZBTB17	137.8	100	100	No OMIM phenotype
ZEB2	163.4	100	99	Mowat-Wilson syndrome, 235730
ZFPM2	196.4	100	99	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	114.5	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 2.12
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors