HEART GENE PANEL DG 2.5/2.6

Gene name	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AARS2	111	99%	97%	Combined oxidative phosphorylation deficiency 8, 614096
ABCC6	94.8	93%	91%	Arterial calcification generalized of infancy 2,614473 Pseudoxanthoma elasticum,264800 Pseudoxanthoma elasticum, forme fruste,177850
ABCC9	156.9	100%	99%	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850
ACAN	99.5	90%	85%	Spondyloepiphyseal dysplasia, Kimberley type, 608361 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800
ACE	106.4	93%	91%	Renal tubular dysgenesis,267430 [Angiotensin I-converting enzyme,benign serum increase] {Alzheimer disease,susceptibility to},104300 {Microvascular complications of diabetes 3},612624 {Myocardial infarction,susceptibility to} {SARS,progression of} {Stroke,hemorrhagic},614519
ACSF3	122.6	100%	97%	Combined malonic and methylmalonic aciduria, 614265
ACTA2	150.7	100%	100%	Aortic aneurysm familial thoracic 6,611788 Moyamoya disease 5,614042 Multisystemic smooth muscle dysfunction syndrome,613834
ACTC1	154.8	100%	100%	Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, familial hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Left ventricular noncompaction 4, 613424
ACTN1	132.9	100%	99%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	143.7	100%	100%	Cardiomyopathy,dilated,1AA,with/without LVNC,612158 Cardiomyopathy,hypertrophic,23,with/without LVNC,612158

ACVR1	169	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ACVR2B	144.8	96%	90%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS6	172.5	100%	99%	No OMIM phenotype Schizophrenia (Fromer (2014) Nature 506,179)
ADAMTS9	143	96%	95%	No OMIM phenotype
ADRB1	116.7	93%	80%	[Resting heart rate],607276 {Congestive heart failure and beta-blocker response,modifier of}
ADRB2	115.7	100%	100%	{Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665 Beta-2-adrenoreceptor agonist, reduced response to
AGL	147.4	100%	99%	Sengers syndrome, 212350 Cataract, autosomal recessive congenital 5, 614691
AGT	182	100%	100%	{Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to} Renal tubular dysgenesis, 267430
AGTR1	137.5	100%	100%	Hypertension, essential, 145500
AKAP9	89.9	97%	92%	Long QT syndrome-11, 611820
ALDH1A2	114.2	100%	100%	No OMIM phenotype Tetralogy of Fallot (Pavan (2009) BMC Med Genet 10, 113) Pentalogy of Cantrell (Steiner (2013) J Med Case Rep 7,287)
ALMS1	167.8	99%	99%	Alstrom syndrome, 203800
ANK2	141.4	99%	99%	Long QT syndrome-4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANKRD1	109.8	97%	93%	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)
ANKS6	77.2	93%	88%	Nephronophthisis 16, 615382
AP1B1	155.5	100%	96%	No OMIM phenotype
AP2B1	116.2	100%	97%	No OMIM phenotype
APBB1	128.5	100%	99%	No OMIM phenotype Dementia alzheimer type,lower risk,association (Hu (1998) Hum Genet 103,295)
ARMC4	123.8	90%	89%	Ciliary dyskinesia, primary, 23, 615451

ATP1A4	157	100%	99%	No OMIM phenotype
BAG3	94.9	100%	99%	Myopathy, myofibrillar, 6, 612954 Cardiomyopathy, dilated, 1HH, 613881
BICC1	161.3	99%	98%	{Renal dysplasia,cystic,susceptibility to},601331
BMPR2	195.7	100%	100%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease, 265450
BRAF	68.9	89%	83%	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707
C1orf127	84.2	99%	96%	No OMIM phenotype Autism (Lim (2013) Neuron 77,235)
C5orf42	119.9	95%	91%	Joubert syndrome 17, 614615
CACNA1B	129.8	94%	89%	?Dystonia 23,614860
CACNA1C	145.4	99%	98%	Timothy syndrome, 601005 Brugada syndrome 3, 611875
CACNA1D	143.9	100%	99%	Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	88.5	93%	86%	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	133.6	100%	97%	Brugada syndrome 4, 611876
CALM1	129.9	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 -3
CALM2	60.4	67%	66%	Long QT syndrome 15,616249

CALM3	114.5	100%	100%	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	138.3	100%	100%	Cardiomyopathy, familial hypertrophic, 19, 613875
CAPN3	113.1	98%	93%	Muscular dystrophy, limb-girdle, type 2A, 253600
CASQ2	148.7	100%	99%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV3	269.3	100%	100%	Muscular dystrophy, limb-girdle, type IC, 607801 Rippling muscle disease, 606072 Creatine phosphokinase, elevated serum, 123320 Myopathy, distal, Tateyama type, 614321 Cardiomyopathy, familial hypertrophic, 192600 Long QT syndrome-9, 6
CBL	118.2	98%	96%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	106.9	99%	91%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D2A	121.8	99%	94%	COACH syndrome,216360 Joubert syndrome 9,612285 Meckel syndrome 6,612284
CCDC151	111.8	100%	95%	Ciliary dyskinesia,primary,30,616037
CCDC39	81.5	95%	90%	Ciliary dyskinesia, primary, 14, 613807
CDKN1C	26.9	59%	53%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CEP290	69.5	89%	76%	?Bardet-Biedl syndrome 14,615991 Joubert syndrome 5,610188 Leber congenital amaurosis 10,611775 Meckel syndrome 4,611134 Senior-Loken syndrome 6,610189
CFC1	47.8	77%	56%	Heterotaxy, visceral, 2, autosomal, 605376 Double-outlet right ventricle, 217095 Transposition of the great arteries, dextro-looped 2, 613853

CHD7	136.1	99%	96%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CITED2	108.7	100%	100%	Ventricular septal defect 2, 614431 Atrial septal defect 8, 614433
CNTF	102.2	100%	100%	No OMIM phenotype Ciliary neurotrophic factor deficiency (Takahashi (1994) Nat Genet 7,79)
CNTRL	111.6	98%	94%	No OMIM phenotype
COL3A1	100.5	89%	84%	Ehlers-Danlos syndrome,type III,130020 Ehlers-Danlos syndrome, type IV,130050
COL4A1	83	96%	89%	Porencephaly 1, 175780
COL5A1	96.6	95%	92%	Ehlers-Danlos syndrome, classic type I,130000
COL5A2	78.4	99%	96%	Ehlers-Danlos syndrome, classic type I,130000
CRELD1	94	98%	96%	{Atrioventricular septal defect, susceptibility to, 2}, 606217 Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRKL	138.5	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	120.2	97%	95%	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184
CSRP3	101	100%	100%	Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, familial hypertrophic, 12, 612124
CTBP2	79.5	98%	93%	No OMIM phenotype Congenital heart disease (Glessner (2014) Circ Res 115,884)
CTF1	24.8	21%	19%	No OMIM phenotype Cardiomyopathy,dilated (Erdmann (2000) Hum Mutat 16,448)

CTLA4	199.3	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	151	100%	100%	Arrhytmogenic right ventricular dysplasia, familial, 13, 615616
CXADR	74.7	92%	87%	No OMIM phenotype
CXCR4	195.6	100%	100%	WHIM syndrome, 193670
CYP11B2	153.5	100%	99%	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Low renin hypertension, susceptibility to Aldosterone to renin ratio raised
DAW1	162.9	100%	92%	No OMIM phenotype
DCTN5	113.4	99%	93%	No OMIM phenotype
DDX39B	20	69%	37%	No OMIM phenotype Leprosy,susceptibility to,association with (Ali (2012) Hum Genet 131,703)
DES	108.8	100%	99%	Arrhytmogenic right ventricular dysplasia, familial, 13, 615616
DMD	83.9	99%	94%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DNAAF3	82.3	97%	89%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	130.3	99%	97%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	123.9	99%	96%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	107.4	98%	96%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAJC19	96.9	99%	88%	3-methylglutaconic aciduria, type V, 610198
DNM2	116	96%	93%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482
DOLK	170.5	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DPP6	132.1	98%	94%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}
DRC1	91.7	100%	98%	Ciliary dyskinesia, primary, 21, 615294
DSC2	145.9	97%	95%	Arrhythmogenic right ventricular dysplasia 11 without/with mild palmoplantar keratoderma and woolly hair,610476

DSG2	128.9	100%	99%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSP	131.6	99%	97%	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
DTNA	160.9	100%	100%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DYNC2H1	91.6	95%	82%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYX1C1	78	96%	84%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EDN1	138.6	100%	100%	auriculocondylar syndrome 3,615706 Question mark ears,isolated,612798 {High density lipoprotein cholesterol level QTL 7}
EDNRA	206.4	100%	100%	mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to},157300
EDNRB	124.7	95%	89%	?{Hirschsprung disease, susceptibility to}, 600155 ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFEMP2	110	100%	100%	Cutis laxa,autosomal recessive,type IB,614437
ELN	91.5	99%	96%	Cutis laxa AD,123700 Supravalvar aortic stenosis,185500
EMD	60	97%	84%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ETS1	117.1	99%	98%	No OMIM phenotype Congenital heart disease (Glessner (2014) Circ Res 115,884)
EYA4	153.1	99%	97%	Deafness, autosomal dominant 10, 601316 Cardiomyopathy, dilated, 1J, 605362
FBN1	161.3	99%	98%	Marfan syndrome, 154700 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Weill-Marchesani syndrome 2, dominant, 608328 Aortic aneurysm, ascending, and dissection Stiff skin syndrome, 184900 Acromicric dysplasia, 102370

FBN2	154.3	99%	99%	Contractural arachnodactyly, congenital, 121050
FHL1	58.5	96%	75%	Emery-Dreifuss muscular dystrophy 6,X-linked,300696 Myopathy,X-linked,with postural muscle atrophy,300696 Reducing body myopathy,X-linked 1a,severe,infantile or early childhood onset,300717 Reducing body myopathy,X-linked 1b,with late childhood or adult onset,300718 Scapuloperoneal myopathy,X-linked dominant,300695
FHL2	142.4	98%	97%	No OMIM phenotype Distal myopathy (Evila (2016) Neuromuscul Disord 26,7) Cardiomyopathy,hypertrophic (Friedrich (2014) Basic Res Cardiol 109,451)
FKTN	137.2	97%	89%	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	85.1	99%	97%	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	149.8	100%	99%	Myopathy, myofibrillar, 5, 609524 Myopathy, distal, 4, 614065
FOXC2	42.2	92%	74%	Lymphedema-distichiasis syndrome with/without renal disease and diabetes mellitus,153400
FOXH1	43.1	89%	72%	No OMIM phenotype Congential heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J Cardiol 145,83)

FOXJ1	58	100%	92%	No OMIM phenotype
FOXL1	50.8	82%	76%	No OMIM phenotype Hypoplastic left heart syndrome (Iascone (2012) Clin Genet 81,542)
FREM2	164.1	99%	98%	Fraser syndrome, 219000
FUZ	87.5	99%	97%	Neural tube defects, 182940
FXN	82	75%	75%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GAA	90.9	100%	99%	Glycogen storage disease II, 232300
GATA4	72.6	63%	58%	?Testicular anomalies with or without congenital heart disease,615542 Atrial septal defect 2,607941 Atrioventricular septal defect 4,614430 Tetralogy of Fallot,187500 Ventricular septal defect 1,614429
GATA5	45.2	98%	94%	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	49.7	83%	66%	Atrioventricular septal defect 5, 614474 Atrial septal defect 9, 614475 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	139.4	99%	92%	Cardiomyopathy, dilated, 2B, 614672
GDF1	15.6	80%	41%	Double-outlet right ventricle, 217095 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854 Right atrial isomerism, 208530

GJA1	205.3	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	225.4	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic, 108770
GJC1	190.5	100%	100%	No OMIM phenotype
GLA	47.8	99%	90%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GPD1L	150.3	100%	98%	Brugada syndrome 2, 611777
GTPBP3	107.7	100%	98%	Combined oxidative phosphorylation deficiency 23
H19				Beckwith-Wiedemann syndrome,130650 Silver-Russell syndrome,180860 Wilms tumor 2,194071
HAND1	72.2	100%	94%	No OMIM phenotype Ventricular septal defect (Cheng (2011) Clin Chim Acta) Cardiac malformations (Reamon-Buettner (2009) Hum Mol Genet 18,3567)
HAND2	39.9	99%	87%	No OMIM phenotype Congenital heart disease (Shen (2010) Chin Med J (Engl) 123,1623) Tetralogy of Fallot (Topf (2014) PLoS One 9,e95453)
HCN1	117.4	100%	100%	Epileptic encephalopathy,early infantile,24,615871
HCN4	80.9	98%	90%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HECTD1	169.3	98%	96%	No OMIM phenotype
HEY2	116.5	97%	91%	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)

HFE	126.5	100%	97%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferrin serum level QTL2],614193
HFE2	106.1	100%	100%	Hemochromatosis type 2A,602390
HOOK1	73.5	96%	82%	No OMIM phenotype
HRAS	165.7	100%	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, somatic}, 162900 {Spitz nevus or nevus spilus,somatic},137550 {Thyroid carcinoma, follicular, somatic}, 188470
IDUA	94.1	89%	85%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015
IFNG	120.7	100%	99%	{AIDS,rapid progression of},609423 {Aplastic anemia},609135 {Hepatitis C virus,response to therapy of},609532 {TSC2 angiomyolipomas,renal,modifier of},613254 {Tuberculosis,protection against},607948
IFT140	105.2	99%	96%	Mainzer-Saldino syndrome, 266920
IFT74	87.4	97%	82%	No OMIM phenotype
IL10	132.2	100%	100%	{Graft-versus-host disease,protection against},614395 {HIV-1,susceptibility to},609423 {Rheumatoid arthritis,progression of},180300
ILK	131.2	100%	100%	No OMIM phenotype Cardiomyopathy, dilated (Knoll (2007) Circulation 116,515)
IRX4	76.9	96%	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	139.8	100%	97%	Alagille syndrome, 118450

JPH2	74.8	96%	82%	Cardiomyopathy, familial hypertrophic 17, 613873
JUP	134.9	100%	99%	Arrhythmogenic right ventricular dysplasia 12,611528 Naxos disease,601214
KCNA5	123.4	99%	96%	Atrial fibrillation, familial, 7, 612240
KCND2	182.2	100%	99%	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	172.2	100%	100%	Spinocerebellar ataxia 19, 607346
KCNE1	443.9	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome-5, 613695
KCNE1L	54.2	86%	74%	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	113.9	100%	100%	Long QT syndrome-6, 613693 Atrial fibrillation, familial, 4, 611493
KCNE3	171.2	100%	100%	Brugada syndrome 6, 613119
KCNE4	85.1	79%	76%	No OMIM phenotype Periodic paralysis (Silva (2004) Arq Bras Endocrinol Metabol 48,196) Atrial fibrillation, association with (Zeng (2007) Cardiology 108,97)
KCNH2	89.2	93%	83%	Long QT syndrome-2, 613688 {Long QT syndrome-2, acquired, susceptibility to}, 613688 Short QT syndrome-1, 609620
KCNJ11	257	100%	100%	Diabetes mellitus,permanent neonatal,with neurologic features,606176 Diabetes mellitus,transient neonatal,3,610582 Hyperinsulinemic hypoglycemia,familial,2,601820 Maturity-onset diabetes of the young,type 13,616329 {Diabetes mellitus,type 2,susceptibility to},125853
KCNJ12	480.2	100%	100%	No OMIM phenotype
KCNJ2	212.3	100%	100%	Andersen syndrome, 170390 Short QT syndrome-3, 609622 Atrial fibrillation, familial, 9, 613980

KCNJ3	162	100%	100%	No OMIM phenotype Schizophrenia, association with (Yamada (2012) Hum Genet 131,443)
KCNJ5	179.7	100%	99%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ8	178.4	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	122.9	100%	100%	{Hypertension,diastolic,resistance to},608622
KCNQ1	96.7	88%	85%	Long QT syndrome-1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Atrial fibrillation, familial, 3, 607554 Short QT syndrome-2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ1OT1				Beckwith-Wiedemann syndrome,130650
KCNQ2	80.5	98%	92%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720
KIF7	70	91%	84%	Hydrolethalus syndrome 2, 614120
KMT2D	133.1	99%	99%	Kabuki syndrome 1, 147920
KRAS	57.8	100%	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-Mins syndrome,somatic mosaic,163200
LAMA4	129	100%	99%	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	76.4	92%	91%	Danon disease, 300257

LDB3	108.4	94%	92%	Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated 1C, 601493 Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493
LEFTY2	36.5	75%	65%	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LIMS1	49.9	41%	34%	No OMIM phenotype
LMNA	72	95%	87%	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	122.8	100%	92%	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	177.9	99%	99%	No OMIM phenotype Keratosis pilaris atrophicans (Klar (2015) J Med Genet 52,599) Abdominal aortic aneurysm, increased risk (Bown (2011) Am J Hum Genet 89,619) Multiple autoimmune syndrome (Johar (2015) J Transl Med 13,173) Aortic aneurysm (Li (2014)
LRP2	176.4	100%	99%	Donnai-Barrow syndrome, 222448
LRP6	164.7	100%	99%	{Coronary artery disease,autosomal dominant, 2},610947
LRRC10	163.8	100%	100%	No OMIM phenotype Cardiomyopathy,dilated (Qu (2015) Mol Med Rep 12,3718)
LTBP1	137.5	93%	93%	No OMIM phenotype Autism (Sanders (2012) Nature 485,237) Pseudoexfoliation syndrome (Schlotzer-Schrehardt (2005) Invest.Opthal. Vis. Sci. 46)

MAP2K1	89.5	100%	98%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	103.9	94%	90%	Cardiofaciocutaneous syndrome 4, 615280
MCTP2	128.3	98%	97%	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	131.9	100%	98%	Transposition of the great arteries, dextro-looped 1, 608808
MEF2C	112.8	99%	88%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443
MEGF8	109	99%	96%	Carpenter syndrome 2, 614976
MIB1	144.8	99%	99%	Left ventricular noncompaction 7, 615092
MICA	27.4	70%	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	14.9	50%	26%	No OMIM phenotype autism, association with (Lim (2013) Neuron 77,235)
MMP21	92.7	87%	83%	Heterotaxy,visceral,7,autosomal,616749
MRPL3	61.9	88%	72%	Combined oxidative phosphorylation deficiency 9, 614582
MTO1	155.5	90%	88%	Combined oxidative phosphorylation deficiency 10, 614702
MYBPC3	129.6	97%	93%	Cardiomyopathy, familial hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYH10	137.6	99%	98%	No OMIM phenotype Intellectual disability (Hamdan (2014) PLoS Genet 10,e1004772) Intrauterine growth restriction,microcephaly,developmental delay and hip dysplasia (Tuzovic (2013) Rare Dis 1,e26144)
MYH11	124.4	99%	98%	Aortic aneurysm, familial thoracic 4, 132900

MYH6	107.5	98%	93%	Cardiomyopathy, familial hypertrophic, 14, 613251 Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 {Sick sinus syndrome 3}, 614090
MYH7	106.3	99%	95%	Cardiomyopathy, familial hypertrophic, 1, 192600 Cardiomyopathy, dilated, 1S, 613426 Myopathy, myosin storage, 608358 Laing distal myopathy, 160500 Scapuloperoneal syndrome, myopathic type, 181430 Left ventricular noncompaction 5, 613426
MYH7B	96.8	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	117.5	98%	83%	Cardiomyopathy, familial hypertrophic, 10, 608758
MYL3	119.4	100%	100%	Cardiomyopathy, familial hypertrophic, 8, 608751
MYL7	142.3	100%	100%	No OMIM phenotype
MYLK	144	99%	99%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	95.4	99%	99%	Cardiomyopathy, hypertrophic, midventricular, digenic, 192600
MYO1C	94.1	98%	97%	No OMIM phenotype Sensorineural hearing loss,bilateral (Zadro (2009) Biochim Biophys Acta 1792,27)
MYOM2	149	100%	98%	No OMIM phenotype Tetralogy of Fallot (Grunert (2014) Hum Mol Genet 23,3115)
MYOT	144.1	100%	96%	Muscular dystrophy, limb-girdle, type 1A, 159000 Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ1	79.8	100%	100%	No OMIM phenotype
MYOZ2	145.5	100%	100%	Cardiomyopathy, familial hypertrophic, 16, 613838
MYPN	147	99%	97%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomypathy, familial hypertrophic, 22, 615248 Cardiomyopaty, familial restrictive 4, 615248
MYZAP	136.9	94%	93%	No OMIM phenotype

NAT8	147.3	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	182	100%	100%	Mental retardation,autosomal recessive 46,616116
NEBL	110.1	97%	92%	No OMIM phenotype Cardiomyopathy,dilated (Purejav (2010) J Am Coll Cardiol 56,1493)
NEK8	158	100%	100%	?Nephronophthisis 9,613824 ?Renal-hepatic-pancreatic dysplasia 2,615415
NEXN	66.2	87%	71%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, familial hypertrophic, 20, 613876
NFATC1	97.7	99%	94%	No OMIM phenotype Tricuspid atresia (Abdul-Sater(2012) PLoS One 7,e49532) Biscuspid aortic valve (Bonachea (2014) BMC Med Genomics 7,56) Congenital heart disease (Glessner (2014) Circ Res 115,884) Tetralogy of Fallot (Silversides (2012) PLoS Genet 8, e1002843) Ventricular septal defect (Zhao (2013) Am J Med Genet A 161,3087)
NFATC4	79.4	95%	95%	No OMIM phenotype Cardiac hypertrophy,protection,association (Poirier (2003) Eur J Hum Genet 11,659
NFKBIL1	8.5	33%	4%	{Rheumatoid arthritis,susceptibility to},180300
NGF	227.1	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NKX2-5	70.2	100%	94%	Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	96.8	100%	98%	Persistent truncus arteriosus, 217095
NODAL	130.9	100%	100%	Heterotaxy, visceral, 5, 270100
NOS1AP	161.6	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	83.8	91%	83%	{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	121.3	99%	97%	Aortic valve disease,109730 Adams-Oliver syndrome 5,616028
NOTCH2	166.5	100%	98%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome,102500
NPPA	78.9	100%	99%	Atrial fibrillation, familial, 6, 612201
NPPB	125.2	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	196.2	100%	92%	Congenital heart defects,multiple types,4,615779
NRAS	179.8	100%	100%	Autoimmune lymphoproliferative syndrome type IV, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500
NSD1	142.2	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
OBSCN	144.5	99%	96%	No OMIM phenotype Cardiomyopathy,dilated (Marston (2015) PLoS One 10,e138568) 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	97.6	89%	80%	Lissencephaly, 607432 Subcortical laminar heterotopia, 607432
PCSK5	151.3	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	99	100%	98%	No OMIM phenotype
PDLIM3	162.7	100%	100%	No OMIM phenotype Cardiomyopathy,dilated (Arola (2007) Mol Genet Metab 90,435 Cardiomyopathy, hypertrophic (Bagnall (2010) Int J Cardiol 145,601)
PITX2	128.1	96%	93%	Axenfeld-Rieger syndrome type 1,180500 Iridogoniodysgenesis,type 2,137600 Peters anomaly,604229 Ring dermoid of cornea,180550
PKD1	23.2	39%	30%	Polycystic kidney disease, adult type I, 173900
PKD1L1	114	99%	98%	No OMIM phenotype Subarachnoid haemorrhage,association with (Yamada (2006) Arterioscler Thromb Vasc Biol 26,1920)
PKP2	90.4	97%	86%	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	117.6	95%	91%	No OMIM phenotype
PLA2G7	123.9	100%	98%	Platelet-activating factor acetylhydrolase deficiency, 614278 Asthma, susceptibility to, 600807 Atopy, susceptibility to, 147050
PLEC	87.5	98%	95%	?Epidermolysis bullosa simplex with nail dystrophy,616487 Epidermolysis bullosa simplex with muscular dystrophy,226670 Epidermolysis bullosa simplex with pyloric atresia,612138 Epidermolysis bullosa simples, Ogna type,131950 Muscular dystrophy,limb-girdle,type 2Q,613723
PLN	163.8	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, familial hypertrophic, 18, 613874
PLXND1	113.1	97%	92%	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	127.5	100%	95%	No OMIM phenotype
PPARGC1A	136.9	100%	99%	No OMIM phenotype Diabetes, type 2, association with (Ek (2001) Diabetologia 44,2220)

PRDM1	130.5	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	131.7	99%	97%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRICKLE1	118.1	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRKAG2	118.2	96%	89%	Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, familial hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740
PRKG1	115.9	96%	90%	Aortic aneurysm, familial thoracic 8, 615436
PSKH1	211.1	100%	100%	No OMIM phenotype
PTK7	141.9	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	55.4	68%	58%	No OMIM phenotype Myopathy,congenital (Muhammad (2013) Hum Mol Genet 22,5229)
PTPN11	93	96%	89%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTPN22	138.3	94%	86%	{Diabetes,type 1,susceptibility to},222100 {Rheumatoid arthritis,susceptibility to},180300 {Systemic lupus erythematosus susceptibility to},152700
PTPRC	99.2	94%	84%	{Hepatitic C virus, susceptibility to}, 609532
PTPRM	147.9	100%	100%	No OMIM phenotype
RAF1	118.6	100%	99%	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RANGRF	80.8	100%	99%	No OMIM phenotype Brugada syndrome (Selga (2015) PLoS One 10,e0132888 Histiocytoid cardiomyopathy (Cataldo (2014)
RBM20	161.8	99%	95%	Cardiomyopathy, dilated, 1DD, 613172

RIT1	154.3	100%	100%	Noonan syndrome 8, 615355
ROBO1	169.1	100%	99%	No OMIM phenotype Developmental dyslexia (Hannula-Jouppi (2005) PLoS Genet 1,e50)
ROBO2	138.7	98%	96%	Vesicoureteral reflux 2, 610878
RPSA	82.1	100%	100%	Asplenia, isolated congenital, 271400
RYR2	134.1	99%	97%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
SCN10A	164	99%	98%	Episodic pain syndrome,familial 2,615551
SCN1B	151.7	100%	96%	Atrial fibrillation,familial,13,615366 Brugada syndrome 5,612838 Cardiac conduction defect,nonspecific,612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2B	176.9	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3B	136.4	100%	100%	Brugada syndrome 7, 613120
SCN4B	67.8	100%	99%	Long QT syndrome-10, 611819
SCN5A	169.5	100%	99%	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy,dilated, 1E,601154 Heart block,nonprogressive,113900 Heart block,progressive,type 1A,113900 Long QT syndrome-3,603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation,familial,1,603829 {Sudden infant death syndrome, susceptibility},272120
SCNN1B	133.8	100%	100%	Bronchiectasis with or without elevated sweat chloride 1,211400 Liddle syndrome, 177200 Pseudohypoaldosteronism,type I,264350
SCNN1G	140.6	98%	96%	Bronchiectasis with or without elevated sweat chloride 3,613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I,264350
SCO2	94.1	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908

SEMA3D	153.4	100%	99%	No OMIM phenotype 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) Congenital heart defects (Sanchez-Castro (2015) Hum Mutat 36,30)
SGCA	136.9	100%	100%	Muscular dystrophy, limb-girdle, type 2D, 608099
SGCB	168.1	96%	95%	Muscular dystrophy, limb-girdle, type 2E, 604286
SGCD	81.4	99%	94%	Muscular dystrophy, limb-girdle, type 2F, 601287 Cardiomyopathy, dilated, 1L, 606685
SGCE	84.1	95%	90%	maternally imprinted Dystonia-11, myoclonic, 159900
SGCG	119.5	100%	100%	Muscular dystrophy, limb-girdle, type 2C, 253700
SHOC2	124.9	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM3	115.1	99%	96%	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	68.1	96%	95%	Shprintzen-Goldberg syndrome, 182212
SLC22A5	137.2	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC25A4	125.7	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418
SLC2A10	138.4	100%	99%	Arterial tortuosity syndrome,208050
SLC8A1	186.3	99%	98%	No OMIM phenotype Colorectal cancer,increased risk,association with (Peters (2012) Hum Genet 131,217) Schizophrenia (Purcell (2014) Nature 506,185)
SLMAP	117.4	91%	81%	No OMIM phenotype Brugada syndrome (Ishikawa (2012) Circ Arrhythm Electrophysiol epub)
SMAD2	152.9	97%	96%	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	112.5	99%	97%	Loeys-Dietz syndrome type 3,613795

SMAD6	78.5	82%	63%	Aortic valve disease 2, 614823
SMARCA4	131.9	99%	94%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609
SMYD1	123.7	100%	100%	No OMIM phenotype
SNTA1	81	80%	73%	Long QT syndrome 12
SNTB1	124.3	100%	97%	No OMIM phenotype
SNX17	139.1	100%	99%	No OMIM phenotype
SOD2	193.3	100%	100%	{Microvascular complications of diabetes 6},612634
SOS1	87.5	93%	88%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733
SUFU	125.3	97%	96%	Medulloblastoma desmoplastic,155255 Basal cell nevus syndrome,109400 {Meningioma,familial,susceptibility to},607174
SYNE1	137.1	99%	98%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	114.9	98%	95%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNPO2	172.8	99%	98%	No OMIM phenotype
TAB1	132.4	99%	97%	No OMIM phenotype
TAB2	189.8	99%	95%	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	74.7	100%	99%	Barth syndrome, 302060
TBC1D32	80.1	97%	91%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
TBX1	70.9	73%	59%	Conotruncal anomaly face syndrome, 217095
TBX20	125.5	100%	99%	Atrial septal defect 4, 611363
TBX3	74.3	99%	94%	Ulnar-mammary syndrome,181450
TBX5	131.6	100%	100%	Holt-Oram syndrome, 142900
TCAP	59.8	100%	100%	Muscular dystrophy, limb-girdle, type 2G, 601954 Cardiomyopathy, dilated, 1N, 607487
TDGF1	137.6	99%	85%	Forebrain defects Forebrain defects (de la Cruz (2002) Hum Genet 110, 422) Congenital heart defects (Roessler (2008) Am J Hum Genet 83, 18)
TFAP2B	117	98%	95%	Char syndrome, 169100
TGFB1	63.6	100%	99%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700

TGFB2	153	100%	100%	Loeys-Dietz syndrome type 4,614816
TGFB3	147.1	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBR1	196.2	93%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	184.5	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 1B, 610168 Loeys-Dietz syndrome, type 2B, 610380
TLL1	146.7	100%	99%	Atrial septal defect 6, 613087
TMEM43	116.8	100%	98%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM67	70.3	91%	84%	COACH syndrome,216360 Joubert syndrome 6,610688 Meckel syndrome 3,607361 Nephronophthisis 11,613550 {Bardet-Biedl syndrome 14,modifier of},209900
TMOD1	122.2	100%	100%	No OMIM phenotype
TMPO	120.1	99%	95%	?Cardiomyopathy,dilated,1T,613740
TNF	13.2	62%	21%	{Asthma,susceptibility to},600807 {Dementia,vascular,susceptibility to} {Malaria,cerebral, susceptibility to},611162 {Migraine without aura,susceptibility to},157300 {Septic shock,susceptibility to}
TNNC1	187.5	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, familial hypertrophic, 13, 613243
TNNI3	89.6	100%	84%	Cardiomyopathy, familial hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 115210 Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	124.6	97%	93%	?Cardiac conduction disease with/without dilated cardiomyopathy,616117

TNNT2	100.7	100%	100%	Cardiomyopathy, familial hypertrophic, 2, 115195 Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TPM1	135.4	100%	97%	Cardiomyopathy, familial hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878 Left ventricular noncompaction 9, 611878
TRDN	68	80%	64%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	127.4	100%	99%	No OMIM phenotype Hypertrophic cardiomyopathy (Chen (2012) Circ Res 111,907)
TRPM4	94.9	99%	94%	Progressive familial heart block, type IB, 604559
TTN	196	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 muscular involvement,603689 Tibial muscular dystrophy,tardive,600334
TTR	138.6	100%	100%	Amyloidosis,hereditary,transthyretin-related,105210 Carpal tunnel syndrome,familial,115430 [Dystransthyretinemic hyperthyroxinemia],145680
VCL	101.4	99%	97%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, familial hypertrophic, 15, 613255
XIRP2	125.4	100%	99%	No OMIM phenotype Schizphrenia (Fromer (2014) Nature 506,179)
ZBTB14	207.1	100%	99%	No OMIM phenotype
ZBTB17	120.3	100%	100%	No OMIM phenotype
ZEB2	149.6	100%	98%	Mowat-Wilson syndrome, 235730
ZFPM2	188.7	100%	100%	Tetralogy of Fallot, 187500 Diaphragmatic hernia 3, 610187
ZIC3	71.1	100%	99%	Heterotaxy, visceral, 1, X-linked 306955 Congenital heart defects, nonsyndromic, 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 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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