HEART GENE PANEL DG 2.15 (282 genes)

Releasedate: 31-01-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ABCC6	116.4	93.6	92.6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	157.9	99.9	99.2	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAD8	141.5	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	135.2	98.4	95.7	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	118.8	98.7	95.1	VLCAD deficiency, 201475
ACSF3	128.8	99.9	99.3	Combined malonic and methylmalonic aciduria, 614265
ACTA2	137.6	100	99.8	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTC1	164.1	100	99.6	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTN2	156.3	100	100	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACVR1	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100
ACVR2B	140.5	97.1	94.7	Heterotaxy, visceral, 4, autosomal, 613751
ADCY5	129.2	92.3	89.1	Dyskinesia, familial, with facial myokymia, 606703
AGK	112.1	99.3	96.4	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146.7	99.7	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	109.5	99	95.1	Lipodystrophy, congenital generalized, type 1, 608594

AKAP9	98	98.3	94.2	?Long QT syndrome-11, 611820
ALDH1A2	114.8	100	99.6	No OMIM phenotype Tetralogy of Fallot (Pavan (2009) BMC Med Genet 10, 113) Pentalogy of Cantrell (Steiner (2013) J Med Case Rep 7,287) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89,476)
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
ALPK3	98.7	94.6	92.5	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	160.3	100	99.9	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKRD1	101.7	99.5	96.8	No OMIM phenotype Cardiomyopathy,hypertrophic (Arimura (2009) J Am Coll Cardiol 54,334) Cardiomyopathy,dilated (Duboscq-Bidot (2009) Eur Heart J 30,2128) ?Total anomalous pulmonary venous return (Cinquetti (2008) Hum Mutat 29,468) ?Neurodevelo
ATPAF2	101.4	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	136.5	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	58.3	98	88.1	Nestor-Guillermo progeria syndrome, 614008
BGN	128.9	100	99.5	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BSCL2	113.5	100	100	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BVES	115	99.8	98.2	?Cardiac arrhythmia with increased serum creatine kinase, 616812
CACNA1C	154.6	99.9	99.2	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	149.9	98	97.8	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	82.7	93.1	84.4	No OMIM phenotype Brugada syndrome (Burashnikov (2010) Heart Rhythm 7,1872) Short QT syndrome (Templin (2011) Eur Heart J 32,1077) Histiocytoid cardiomyopathy (Cataldo (2014) Cardiol Young epub)

				West syndrome (Hino-Fukuyo (2015) Hum Genet 134,
CACNB2	150.9	99.5	96.9	Brugada syndrome 4, 611876
CALM1	114.3	100	99.7	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	54	67.8	65.8	Long QT syndrome 15, 616249
CALM3	117.4	99.9	99.5	No OMIM phenotype Catecholaminergic polymorphic ventricular tachycardia (Boczek (2013) Circulation 128,A14699) Long QT syndrome (Reed (2015) Heart Rhythm 12,419) {Cardiomyopathy,hypertrophic,modifier of} (Friedrich (2009) Eur Heart J 30,1648)
CASQ2	143.3	99.9	99.2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV1	265.4	100	100	?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 Pulmonary hypertension, primary, 3, 615343
CAV3	304.7	100	100	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072
CAVIN4	159.5	100	100	No OMIM phenotype
CDH2	137.5	98.4	97.5	No OMIM phenotype
CFAP53	146.6	97.6	94.2	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	74.5	82.7	71.3	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	142.7	100	99.8	No OMIM phenotype
CITED2	111.6	99.2	99	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	104.3	97.8	92.3	Ehlers-Danlos syndrome, vascular type, 130050

COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COX15	98.6	100	99.7	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
CPT1A	169.3	100	98.7	CPT deficiency, hepatic, type IA, 255120
CPT2	162.8	97.2	95.4	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CRELD1	114.4	99.9	97.8	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRKL	166.4	100	99.8	No OMIM phenotype ?Congenital heart defect (Breckpot (2012) Am J Med Genet A 158A,574) ?Tetralogy of Fallot (Tomita-Mitchell (2012) Physiol Genomics 44,518) ?Ventricular septal defect (Zhao (2013) Am J Med Genet A 161,3087
CRYAB	125.7	99.9	98.7	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CSRP3	103	100	99.9	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CTF1	24.5	28	20	No OMIM phenotype Cardiomyopathy,dilated (Erdmann (2000) Hum Mutat 16,448)
CTNNA3	138.3	100	99.9	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
DES	120.8	99.9	98.1	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DMD	112.4	99.4	97.4	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DOLK	202.9	100	99.9	Congenital disorder of glycosylation, type Im, 610768
DPM3	183.9	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937

DPP6	145.5	96.5	94.5	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	128.5	99.4	96.2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSG2	140.6	99.9	98.7	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSP	154	100	99.8	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DTNA	156.5	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
EEF1A2	177.7	98.8	93.8	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP2	120.9	100	99.9	Cutis laxa, autosomal recessive, type IB, 614437
ELN	91.1	99.4	97.4	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
EMD	100.3	99.8	97.2	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMILIN1	75.6	96.8	87.5	No OMIM phenotype Connective tissue disease, autosomal dominant (Capuano (2016) Hum Mutat 37, 84)
ENPP1	134.8	92.4	83.2	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
EYA4	160.6	100	99.5	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FAH	151.3	100	99.9	Tyrosinemia, type I, 276700

FBN1	159.8	99.9	99.5	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FBN2	161.7	100	99.5	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXO32	150.6	100	100	No OMIM phenotype
FGF12	95.5	99.6	96.3	Epileptic encephalopathy, early infantile, 47, 617166
FHL1	87.2	98.8	93	?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy, X-linked dominant, 300695
FHL2	149.6	99.6	98.4	No OMIM phenotype Cardiomyopathy,hypertrophic (Friedrich (2014) Basic Res Cardiol 109,451) ?Distal myopathy (Evila (2016) Neuromuscul Disord 26,7)
FHOD3	135.2	99.9	98.5	No OMIM phenotype
FKRP	94.5	100	99.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	120	99.2	94.2	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588

FLNA	138.1	100	99.5	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNC	165	100	99.7	Cardiomyopathy, familial hypertrophic, 26, 0 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLT4	155.9	98.6	97.9	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FOXD4	3.1	25	13.3	No OMIM phenotype
FOXE3	20.6	69	47.8	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXH1	47.2	98.5	85	No OMIM phenotype Congenital heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J Cardiol 145,83)
GAA	128.5	100	99.9	Glycogen storage disease II, 232300
GATA4	87.4	68.6	60.7	?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	44.2	98.3	84.4	Congenital heart defects, multiple types, 5, 617912
GATA6	61.7	83.7	72.1	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500

GATAD1	126.6	97	92	?Cardiomyopathy, dilated, 2B, 614672
GBE1	145.5	99.6	97.2	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDF1	19.5	65	48.4	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF2	163.2	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GJA5	268.4	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	81.3	99.7	97.6	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GMPPB	228.5	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNPTAB	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GPD1L	138.3	100	98.5	Brugada syndrome 2, 611777
HADHA	84.4	96.5	90.3	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	80.5	92.5	79.5	Trifunctional protein deficiency, 609015
HAND1	84.8	100	98.9	No OMIM phenotype Ventricular septal defect (Cheng (2011) Clin Chim Acta) Cardiac malformations (Reamon-Buettner (2009) Hum Mol Genet 18,3567) Cardiomyopathy, dilated (Zhou (2015) Clin Chem Lab Med Epub, epub)
HAND2	32.2	87.8	67.9	No OMIM phenotype Tetralogy of Fallot (Topf (2014) PLoS One 9,e95453) Ventricular septal defect (Sun (2016) G3 (Bethesda) epub,epub) ?Congenital heart disease (Shen (2010) Chin Med J (Engl) 123,1623)

HCN2	53.2	58.3	50.7	No OMIM phenotype
HCN3	146.4	99.9	99.2	No OMIM phenotype
HCN4	79.4	98.3	91.8	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HEY2	146.1	99.2	92.8	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
HFE	142	100	99.7	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	116.8	100	100	Hemochromatosis, type 2A, 602390
HSPB6	63.1	89.4	77.4	No OMIM phenotype
IDUA	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
ILK	171.6	100	100	No OMIM phenotype Cardiomyopathy, dilated (Knoll (2007) Circulation 116,515) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89, 476)
IRX3	73.8	82.9	66.2	No OMIM phenotype
IRX4	90	95.8	92.3	No OMIM phenotype Congenital heart defect (Cheng (2014) BMC Genomics 15,1127) {Prostate cancer,susceptibility to} (Nguyen (2012) Hum Mol Genet 21,2076)
ITPA	120.2	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAG1	148.4	98.1	97.5	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JPH2	87.5	90.4	75.1	Cardiomyopathy, hypertrophic, 17, 613873
JUP	145.1	100	99.6	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KCNA5	143.3	99.4	96	Atrial fibrillation, familial, 7, 612240

KCND2	183.4	100	100	No OMIM phenotype Autism and epilepsy (Lee (2014) Hum Mol Genet 23,3481) J-wave syndrome with sudden cardiac death (Perrin (2014) Circ Cardiovasc Genet 7,782) Epilepsy,temporal lobe (Singh (2006) Neurobiol Dis 24,245)
KCND3	182.5	99.9	99.1	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	462.6	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	181.5	100	100	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	177.1	100	100	?Brugada syndrome 6, 613119
KCNE4	84.8	79.9	77.6	No OMIM phenotype ?Periodic paralysis (Silva (2004) Arq Bras Endocrinol Metabol 48,196) {Atrial fibrillation, association with} (Zeng (2007) Cardiology 108,97)
KCNE5	88	97.7	90.3	No OMIM phenotype Atrial fibrillation (Ravn (2008) Heart Rhythm 5,427 Idiopathic ventricular fibrillation (Ohno (2011) Circ Arrhythm Electrophysiol 4,352) Atrial fibrillation,lone,early-onset (Olesen (2014) Heart Rhythm 11,246)
KCNH2	102.6	92.3	84.8	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ11	299.5	100	100	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ2	229.3	100	100	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ5	193.9	100	99.8	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ8	177.1	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	165.3	98.9	96.1	Pulmonary hypertension, primary, 4, 615344
KCNN3	151.1	100	99.9	No OMIM phenotype
KCNQ1	114.7	93	90.3	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500
KLF10	141.4	100	99.7	No OMIM phenotype
KRAS	64.7	99.9	98.7	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LAMA2	143.5	99.9	99.5	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMA4	132.6	100	99.9	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	106.1	92.7	91.2	Danon disease, 300257
LDB3	127.3	95.5	93.7	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LEFTY2	42.3	91.3	77.1	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LIMS2	110.8	93	92.3	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827

LMNA	89.2	97.9	91.3	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMOD1	177.4	100	100	No OMIM phenotype Megacystis-microcolon-intestinal hypoperistalsis syndrome (Halim (2017) Proc Natl Acad Sci USA 114)
LOX	104.4	99.8	97.6	Aortic aneurysm, familial thoracic 10, 617168
LRIT3	142.4	94.4	94.1	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRRC10	193.6	100	100	No OMIM phenotype Cardiomyopathy,dilated (Qu (2015) Mol Med Rep 12,3718)
LZTR1	134	100	99.4	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MAT2A	115.4	99.7	96.9	No OMIM phenotype Thoracic aortic aneurysms (Guo (2015) Am J Hum Genet 96, 170)
MED13L	134.6	100	99.6	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MFAP5	126.8	100	99.5	Aortic aneurysm, familial thoracic 9, 616166
MIB1	141.7	100	99.6	Left ventricular noncompaction 7, 615092
MLYCD	75.6	91.3	86.9	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	93.3	90.2	84.6	Heterotaxy, visceral, 7, autosomal, 616749
MYBPC3	142.5	98.5	95.7	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYBPHL	99.8	99.2	94.2	No OMIM phenotype
MYH11	132.6	100	99.3	Aortic aneurysm, familial thoracic 4, 132900

MYH6	113.3	99	96.1	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	111.4	99.4	96.8	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYH7B	113.2	97.6	94.5	No OMIM phenotype ?Cardiomyopathy,left ventricular noncompaction (Esposito (2013) Orphanet J Rare Dis 8) ?Hearing loss (Haraksingh (2014) BMC Genomics 15,1155)
MYL2	134.6	98.7	90.1	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	103.1	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	158.5	100	100	?Atrial fibrillation, familial, 18, 617280
MYL7	130.8	100	100	No OMIM phenotype
MYLK	148.4	99.9	99.3	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	120	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYO6	89.7	98.1	92.3	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYOM1	149.5	99.8	98.4	No OMIM phenotype
MYOT	139.4	99.3	95.5	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	145.9	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	142.4	99.3	98.4	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
NCOA6	150.2	100	99.6	No OMIM phenotype
NEBL	102.2	96.7	92.9	No OMIM phenotype Cardiomyopathy,dilated (Purejav (2010) J Am Coll Cardiol 56,1493)

NEXN	79.8	94.2	79.9	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NKX2-5	83.2	100	99.5	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	104.4	100	99.7	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NNT	136.9	98.6	97.1	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NODAL	160.7	100	99.9	Heterotaxy, visceral, 5, 270100
NOS1AP	192.2	100	100	No OMIM phenotype Long QT syndrome (Shigemizu (2015) PLoS One 10,e0130329) ?Obsessive-compulsive disorder (Delorme (2010) BMC Med Genet 11,108) {Cardiac repolarisation, association with} (Arking (2006) Nat Genet 38,644)
NOTCH1	137.5	99.1	98	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	172.4	100	99.9	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPA	115.4	100	100	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPPB	160.4	100	100	No OMIM phenotype ?Hypertension (Zeng (2013) J Hum Hypertens 27,271) {Diabetes type 2,reduced risk,association with} (Meirhaeghe (2007) Hum Mol Genet 16,1343)
NR2F2	246.1	98.7	94.3	Congenital heart defects, multiple types, 4, 615779
NRAS	188.4	100	100	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200

				Thyroid carcinoma, follicular, somatic, 188470
NUP155	115.1	97.6	92.3	?Atrial fibrillation 15, 615770
OBSCN	159.3	99.3	98.2	No OMIM phenotype Cardiomyopathy,dilated (Marston (2015) PLoS One 10,e138568) Glioblastoma (Balakrishnan (2007) Cancer Res 67,3545) ?Breast cancer (Aloraifi (2015) FEBS J epub,epub) ?Schizophrenia (Fromer (2014) Nature 506,179) ?Cardiomyopa
PCCA	103.1	96.4	89.2	Propionicacidemia, 606054
PCCB	129.8	98.7	96.4	Propionicacidemia, 606054
PDLIM3	148.4	100	100	No OMIM phenotype Cardiomyopathy,dilated (Arola (2007) Mol Genet Metab 90,435 ?Cardiomyopathy, hypertrophic (Bagnall (2010) Int J Cardiol 145,601)
PGM1	133.6	100	99.9	Congenital disorder of glycosylation, type It, 614921
PHKA1	106.7	98.9	95.3	Muscle glycogenosis, 300559
PHYH	74.6	97.5	90.8	Refsum disease, 266500
PITX2	147.8	99.7	97.5	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PKD1L1	123.8	100	99.6	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	99.6	94.6	87.7	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	138.9	99.2	96.2	No OMIM phenotype
PLEKHM2	112.7	100	99.7	No OMIM phenotype Cardiomyopathy, dilated with left ventricular noncompaction (Muhammad (2015) Hum Mol Genet 24, 7227)
PLN	209.7	100	100	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	137.9	99.8	97.5	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065

PNPLA2	113.2	99.7	97.4	Neutral lipid storage disease with myopathy, 610717
POMT1	155.7	99.7	98.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	111.1	98.9	97.5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PPA2	80.4	94.6	82.5	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	108.7	99.9	98.4	Cardiomyopathy, dilated, 2C, 618189
PRDM16	161.5	100	99.1	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRKAG2	125.6	98.1	91.6	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKG1	123.4	98.7	95.4	Aortic aneurysm, familial thoracic 8, 615436
PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAF1	127.3	100	99.7	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RANGRF	114	99.9	98.4	No OMIM phenotype Brugada syndrome (Selga (2015) PLoS One 10,e0132888 Histiocytoid cardiomyopathy (Cataldo (2014)
RBM20	180.9	99.2	96.6	Cardiomyopathy, dilated, 1DD, 613172
RIT1	165.6	100	100	Noonan syndrome 8, 615355
RYR2	142.2	99.7	98.4	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
SCN10A	165.3	100	99.5	Episodic pain syndrome, familial, 2, 615551

SCN1B	168.3	97.1	96.1	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN2B	185.8	100	100	Atrial fibrillation, familial, 14, 615378
SCN3B	147.3	100	100	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	77.5	100	97.9	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	169.4	99	99	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 {Sudden infant death syndrome, susceptibility to}, 272120
SDHA	122.2	84.8	80.8	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SGCA	147.3	100	99.7	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	154.2	96.6	94.2	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94.8	100	99.4	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	138.7	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHOC2	140.4	100	99.4	Noonan-like syndrome with loose anagen hair, 607721
SHROOM3	137.3	99.9	98.9	No OMIM phenotype Heterotaxy (Tariq (2011) Genome Biol 12,R91) ?Neural tube defects (Lemay (2015) J Med Genet 52,493)

				{Leukaemia risk,association with} (Rudd (2006) Blood 108,638)
SKI	85.3	96.4	90.8	Shprintzen-Goldberg syndrome, 182212
SLC22A5	153.3	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A20	110.3	100	99.7	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	134.1	100	100	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC2A10	166.4	97.7	97.6	Arterial tortuosity syndrome, 208050
SLMAP	121.2	93.5	85.2	No OMIM phenotype Brugada syndrome (Ishikawa (2012) Circ Arrhythm Electrophysiol epub)
SMAD1	184	99.9	99.7	No OMIM phenotype
SMAD2	151.7	99.9	99.1	No OMIM phenotype Congenital heart disease (Zaidi (2013) Nature 498,220) Arterial aneurysms and dissections (Micha (2015) Hum Mutat 36,1145) Holoprosencephaly (Roessler (2008) Am J Hum Genet 83,18)
SMAD3	131.7	99.9	99.2	Loeys-Dietz syndrome 3, 613795
SMAD4	125.5	100	100	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	100.5	80	72	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMAD9	132.8	100	100	Pulmonary hypertension, primary, 2, 615342
SNTA1	97	82.3	77.3	Long QT syndrome 12, 612955
SOS1	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SRI	114.3	97.9	88.9	No OMIM phenotype
SYNE1	136.6	98.2	97.6	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743

SYNE2	123.1	98.6	96	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
TAB2	210.5	99.7	97.6	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	94	99.9	98.8	Barth syndrome, 302060
TBX1	75.3	77.1	67.4	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX20	142.8	99.9	99.3	Atrial septal defect 4, 611363
TBX5	141.3	100	100	Holt-Oram syndrome, 142900
TCAP	89	100	99.2	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TDGF1	151.4	99.8	96.4	Forebrain defects, 0
TECRL	59.3	89.9	77.1	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	153.5	98.8	96.3	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TGFB2	176.9	100	99.9	Loeys-Dietz syndrome 4, 614816
TGFB3	171.5	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	173.4	93.7	93.6	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	193.5	100	99.9	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TLL1	140.1	100	99.9	Atrial septal defect 6, 613087
TMEM43	124.9	100	99.5	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMPO	117.8	98.7	94.5	?Cardiomyopathy, dilated, 1T, 613740
TNNC1	174.5	100	100	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	86.7	98.1	86.5	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690

TNNI3K	118.8	98.8	96	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	106.3	100	99.9	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TOR1AIP1	143.8	97.6	95.9	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TPM1	132.9	99.7	97.9	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TRDN	71.9	83.6	70.8	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	118.6	100	99.6	No OMIM phenotype Hypertrophic cardiomyopathy (Chen (2012) Circ Res 111,907)
TRPM4	109.2	99.8	98.5	Progressive familial heart block, type IB, 604559
TTN	187.8	98.2	97.2	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334
TTR	152.3	94.6	94.6	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TXNRD2	119.3	93.3	91.2	?Glucocorticoid deficiency 5, 617825
VCL	115.8	100	99.8	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	138.5	100	99.7	No OMIM phenotype ?Schizphrenia (Fromer (2014) Nature 506,179)
XK	96.8	99.9	99.1	McLeod syndrome with or without chronic granulomatous disease, 300842
ZBTB17	142	100	100	No OMIM phenotype
ZFPM2	196.3	100	99.6	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500

ZIC3	113.7	100	99.8	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 31st, 2018.
This list is accurate for panel version DG 2.15
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors