HEART GENE PANEL DG 2.17 (301 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	135.3	100.0%	100.0%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCC6	116.6	93.7%	93.1%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC9	140.4	100.0%	99.9%	Hypertrichotic osteochondrodysplasia, 239850 Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABL1	170.5	100.0%	100.0%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACAD8	131.8	100.0%	99.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	130.9	100.0%	98.8%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	125.2	99.9%	98.7%	VLCAD deficiency, 201475
ACSF3	158.9	99.9%	99.4%	Combined malonic and methylmalonic aciduria, 614265
ACTA2	89.6	99.9%	98.6%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTC1	118.4	100.0%	99.3%	Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098
ACTN2	134.8	100.0%	99.9%	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654
ACVR2B	124.2	99.9%	98.3%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS19	116.8	98.7%	95.0%	No OMIM Disease ID
ADCY5	144.1	98.7%	96.4%	Dyskinesia, familial, with facial myokymia, 606703
AGK	109.6	99.6%	95.5%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691

AGL	141.9	100.0%	99.7%	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGPAT2	180.5	99.7%	97.0%	Lipodystrophy, congenital generalized, type 1, 608594
AKAP9	96.2	99.0%	96.3%	?Long QT syndrome 11, 611820
ALDH1A2	108.8	99.9%	99.0%	No OMIM Disease ID
ALMS1	178.0	100.0%	99.8%	Alstrom syndrome, 203800
ALPK3	126.5	99.7%	97.9%	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	143.6	100.0%	100.0%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKRD1	98.9	99.9%	98.6%	No OMIM Disease ID
ARIH1	119.6	100.0%	99.3%	No OMIM Disease ID
ATPAF2	109.2	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	189.4	100.0%	99.9%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	52.9	97.3%	86.5%	Nestor-Guillermo progeria syndrome, 614008
BGN	147.7	100.0%	100.0%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BRAF	71.0	91.7%	79.4%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BSCL2	112.9	100.0%	99.9%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
BVES	109.4	100.0%	98.4%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
CACNA1C	151.7	99.9%	99.4%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	135.3	98.0%	97.8%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	91.1	98.0%	92.3%	No OMIM Disease ID
CACNB2	135.8	100.0%	99.4%	Brugada syndrome 4, 611876
CALM1	95.2	99.8%	98.1%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916

CALM2	43.0	66.5%	61.9%	Long QT syndrome 15, 616249
CALM3	105.3	100.0%	99.9%	No OMIM Disease ID
CASQ2	113.9	100.0%	99.3%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV1	200.0	100.0%	100.0%	Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526
CAV3	233.9	100.0%	100.0%	Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072 Cardiomyopathy, familial hypertrophic, 192600
CDH2	113.6	99.9%	98.9%	No OMIM Disease ID
CFAP53	135.5	99.2%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	136.1	91.1%	82.3%	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	143.6	100.0%	99.5%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	119.4	100.0%	99.9%	No OMIM Disease ID
CITED2	174.9	99.2%	99.2%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	99.6	99.4%	97.0%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COQ2	107.7	97.7%	97.0%	Coenzyme Q10 deficiency, primary, 1, 607426
COX15	90.4	99.9%	98.7%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
CPT1A	135.4	99.9%	98.6%	CPT deficiency, hepatic, type IA, 255120
CPT2	152.7	98.3%	98.3%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CRELD1	105.6	99.9%	96.0%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRYAB	96.3	99.9%	97.9%	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CSRP3	94.3	99.9%	97.8%	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CTNNA3	128.9	100.0%	99.9%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616

DES	138.0	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DMD	107.5	99.6%	97.9%	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DOLK	171.4	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DPM3	218.6	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	129.8	100.0%	99.8%	Mental retardation, autosomal dominant 33, 616311
DSC2	120.1	99.7%	97.3%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	131.5	100.0%	99.1%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSP	148.0	100.0%	99.6%	Keratosis palmoplantaris striata II, 612908 Epidermolysis bullosa, lethal acantholytic, 609638 Skin fragility-woolly hair syndrome, 607655 Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DTNA	127.7	100.0%	99.9%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
EEF1A2	209.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP2	141.3	100.0%	100.0%	Cutis laxa, autosomal recessive, type IB, 614437
EHMT1	138.4	94.7%	94.5%	Kleefstra syndrome 1, 610253
ELN	113.2	100.0%	99.6%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
EMD	147.1	99.9%	99.1%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMILIN1	126.9	99.9%	99.0%	No OMIM Disease ID
ENPP1	128.6	97.9%	92.4%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
EYA4	134.5	100.0%	100.0%	Deafness, autosomal dominant 10, 601316 ?Cardiomyopathy, dilated, 1J, 605362
FAH	136.7	100.0%	99.8%	Tyrosinemia, type I, 276700
FBN1	138.3	100.0%	99.7%	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308

				Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FBN2	144.5	100.0%	99.9%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXO32	143.6	100.0%	100.0%	No OMIM Disease ID
FGF12	105.7	100.0%	100.0%	Epileptic encephalopathy, early infantile, 47, 617166
FHL1	68.2	99.2%	92.8%	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696
FHL2	169.2	99.9%	99.3%	No OMIM Disease ID
FHOD3	141.0	100.0%	99.4%	No OMIM Disease ID
FKRP	178.0	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	108.0	99.9%	96.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLNA	156.4	100.0%	99.9%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620

FLNC	169.4	100.0%	99.9%	Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Cardiomyopathy, familial hypertrophic, 26, 0
FLT4	177.8	99.2%	99.2%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FOXC2	144.2	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	6.0	36.3%	21.3%	No OMIM Disease ID
FOXE3	111.7	93.8%	87.3%	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256
FOXH1	96.2	100.0%	100.0%	No OMIM Disease ID
FOXL1	167.0	100.0%	100.0%	No OMIM Disease ID
GAA	180.3	100.0%	100.0%	Glycogen storage disease II, 232300
GATA4	95.7	98.9%	90.6%	?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429
GATA5	84.0	100.0%	99.5%	Congenital heart defects, multiple types, 5, 617912
GATA6	128.0	99.6%	95.4%	Pancreatic agenesis and congenital heart defects, 600001 Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	143.2	100.0%	100.0%	?Cardiomyopathy, dilated, 2B, 614672
GATB	102.6	100.0%	99.9%	No OMIM Disease ID
GATC	153.3	100.0%	100.0%	No OMIM Disease ID
GBE1	152.5	100.0%	99.5%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GDF1	62.1	99.4%	91.2%	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GDF2	157.3	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GJA5	225.2	100.0%	100.0%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	74.4	99.4%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500

GLB1	87.4	99.5%	95.2%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GMPPB	233.1	100.0%	100.0%	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
GNB2	223.0	100.0%	100.0%	No OMIM Disease ID
GNPTAB	149.5	100.0%	99.4%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GPD1L	132.4	100.0%	99.9%	Brugada syndrome 2, 611777
HADHA	74.6	96.1%	89.6%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HADHB	76.9	96.0%	83.7%	Trifunctional protein deficiency, 609015
HAND1	189.1	100.0%	100.0%	No OMIM Disease ID
HAND2	97.6	100.0%	99.9%	No OMIM Disease ID
HCN2	41.0	71.2%	58.2%	No OMIM Disease ID
HCN3	159.3	100.0%	99.8%	No OMIM Disease ID
HCN4	109.8	100.0%	99.9%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HEY2	179.4	99.4%	95.6%	No OMIM Disease ID
HFE	114.7	100.0%	99.1%	Hemochromatosis, 235200
HFE2	162.3	100.0%	100.0%	Hemochromatosis, type 2A, 602390
HSPB6	112.2	98.4%	91.7%	No OMIM Disease ID
IDUA	169.2	99.3%	96.4%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
ILK	133.9	100.0%	99.8%	No OMIM Disease ID
ITPA	142.5	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647
JAG1	143.4	99.4%	97.6%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
JPH2	139.7	99.7%	97.8%	Cardiomyopathy, hypertrophic, 17, 613873

JUP	137.0	100.0%	99.9%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KCNA5	173.9	100.0%	100.0%	Atrial fibrillation, familial, 7, 612240
KCND2	166.7	100.0%	100.0%	No OMIM Disease ID
KCND3	180.1	99.9%	99.3%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	398.8	100.0%	100.0%	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNE2	127.8	100.0%	98.4%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	149.5	100.0%	100.0%	?Brugada syndrome 6, 613119
KCNE4	125.2	80.5%	80.5%	No OMIM Disease ID
KCNE5	142.8	100.0%	99.7%	No OMIM Disease ID
KCNH2	120.8	99.0%	96.3%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620
KCNJ11	222.1	100.0%	100.0%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ2	164.2	100.0%	100.0%	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KCNJ5	171.5	100.0%	100.0%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ8	126.3	100.0%	100.0%	No OMIM Disease ID
KCNK3	184.4	100.0%	99.7%	Pulmonary hypertension, primary, 4, 615344
KCNN3	130.4	100.0%	99.9%	Zimmermann-Laband syndrome 3, 618658
KCNQ1	150.5	98.9%	96.5%	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554
KLF10	137.2	100.0%	99.9%	No OMIM Disease ID
KLHL24	174.7	100.0%	100.0%	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KMT2D	150.7	100.0%	99.9%	Kabuki syndrome 1, 147920
KRAS	64.0	99.8%	96.8%	Leukemia, acute myeloid, 601626 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470

				Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
LAMA2	131.6	100.0%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA4	118.6	100.0%	99.8%	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	89.8	97.8%	92.3%	Danon disease, 300257
LDB3	161.1	96.3%	95.0%	Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493
LEFTY2	77.2	99.6%	94.4%	No OMIM Disease ID
LIMS2	131.0	96.5%	93.4%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMNA	118.2	98.3%	93.2%	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMOD1	166.0	100.0%	100.0%	No OMIM Disease ID
LOX	176.0	100.0%	99.1%	Aortic aneurysm, familial thoracic 10, 617168
LRRC10	196.4	100.0%	100.0%	No OMIM Disease ID
LTBP3	166.1	100.0%	100.0%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	157.2	100.0%	99.9%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAT2A	96.6	99.9%	96.5%	No OMIM Disease ID
MCTP2	120.7	99.5%	97.5%	No OMIM Disease ID

MED13L	112.2	100.0%	99.8%	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MFAP5	109.5	99.8%	95.3%	Aortic aneurysm, familial thoracic 9, 616166
MIB1	129.1	100.0%	100.0%	Left ventricular noncompaction 7, 615092
MLYCD	105.5	99.7%	97.3%	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	103.7	100.0%	99.5%	Heterotaxy, visceral, 7, autosomal, 616749
MYBPC3	154.3	100.0%	98.9%	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYBPHL	87.4	99.5%	96.8%	No OMIM Disease ID
MYH11	131.1	100.0%	99.7%	Aortic aneurysm, familial thoracic 4, 132900
MYH6	104.2	99.3%	96.3%	Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 Cardiomyopathy, dilated, 1EE, 613252
MYH7	101.7	99.7%	97.2%	Myopathy, myosin storage, autosomal recessive, 255160 Left ventricular noncompaction 5, 613426 Laing distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Cardiomyopathy, hypertrophic, 1, 192600
MYH7B	132.0	99.9%	97.6%	No OMIM Disease ID
MYL2	139.8	99.9%	97.0%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	106.8	100.0%	100.0%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	139.8	100.0%	100.0%	?Atrial fibrillation, familial, 18, 617280
MYL7	146.5	100.0%	100.0%	No OMIM Disease ID
MYLK	132.8	100.0%	99.6%	Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210
MYLK2	141.9	100.0%	100.0%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	138.3	99.8%	98.5%	No OMIM Disease ID
MYO6	98.8	99.5%	95.6%	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYOM1	125.2	99.9%	98.9%	No OMIM Disease ID
MYOT	142.2	100.0%	99.1%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	142.3	100.0%	100.0%	Cardiomyopathy, hypertrophic, 16, 613838

MYPN	128.9	99.9%	99.0%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
NCOA6	133.9	100.0%	99.9%	No OMIM Disease ID
NEBL	98.4	98.2%	95.8%	No OMIM Disease ID
NEXN	87.7	96.0%	85.4%	Cardiomyopathy, hypertrophic, 20, 613876 Cardiomyopathy, dilated, 1CC, 613122
NKX2-5	141.9	100.0%	100.0%	Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	159.7	100.0%	100.0%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NNT	127.0	99.9%	98.1%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NODAL	156.2	100.0%	100.0%	Heterotaxy, visceral, 5, 270100
NOS1AP	213.9	100.0%	100.0%	No OMIM Disease ID
NOTCH1	158.4	99.8%	99.2%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	130.5	100.0%	99.8%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPPA	172.3	100.0%	100.0%	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPPB	238.4	100.0%	100.0%	No OMIM Disease ID
NR2F2	262.6	100.0%	100.0%	Congenital heart defects, multiple types, 4, 615779
NRAS	145.3	100.0%	100.0%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NUP155	117.6	98.5%	96.6%	?Atrial fibrillation 15, 615770
OBSCN	165.9	100.0%	99.6%	No OMIM Disease ID
PCCA	97.7	99.1%	95.4%	Propionicacidemia, 606054

PCCB	114.9	99.5%	97.1%	Propionicacidemia, 606054
PDLIM3	150.7	100.0%	99.9%	No OMIM Disease ID
PDLIM5	123.3	92.1%	88.9%	No OMIM Disease ID
PEX5	115.8	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGM1	134.8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PHKA1	90.2	97.8%	91.7%	Muscle glycogenosis, 300559
PHYH	75.9	100.0%	97.9%	Refsum disease, 266500
PITX2	186.2	100.0%	99.6%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PKD1L1	113.1	100.0%	99.5%	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	97.3	97.4%	91.6%	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	122.6	99.8%	97.3%	No OMIM Disease ID
PLD1	116.6	100.0%	99.6%	Cardiac valvular defect, developmental, 212093
PLEKHM2	144.5	100.0%	99.9%	No OMIM Disease ID
PLN	150.9	100.0%	100.0%	Cardiomyopathy, hypertrophic, 18, 613874 Cardiomyopathy, dilated, 1P, 609909
PLOD1	141.5	99.9%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PMEPA1	128.3	100.0%	99.9%	No OMIM Disease ID
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	159.9	100.0%	99.9%	Neutral lipid storage disease with myopathy, 610717
POMT1	137.5	99.6%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	109.7	100.0%	99.1%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156
PPA2	92.7	98.0%	90.2%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	166.9	100.0%	99.2%	Cardiomyopathy, dilated, 2C, 618189

PRDM16	230.2	100.0%	99.3%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRKAG2	135.6	99.1%	96.5%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKG1	127.2	99.8%	98.8%	Aortic aneurysm, familial thoracic 8, 615436
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
QRSL1	86.0	98.8%	93.2%	No OMIM Disease ID
RAF1	111.1	100.0%	99.9%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RANGRF	154.1	100.0%	99.5%	No OMIM Disease ID
RBM20	193.1	100.0%	99.8%	Cardiomyopathy, dilated, 1DD, 613172
RIT1	142.5	100.0%	100.0%	Noonan syndrome 8, 615355
RRAD	157.7	96.8%	91.4%	No OMIM Disease ID
RRAGC	92.3	100.0%	99.5%	No OMIM Disease ID
RYR2	127.4	99.9%	98.7%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
SCN10A	141.5	100.0%	99.5%	Episodic pain syndrome, familial, 2, 615551
SCN1B	186.5	100.0%	99.3%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2B	190.3	100.0%	100.0%	Atrial fibrillation, familial, 14, 615378
SCN3B	145.5	100.0%	100.0%	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120
SCN4B	68.2	99.9%	98.0%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN5A	154.0	99.0%	99.0%	Atrial fibrillation, familial, 10, 614022 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 Long QT syndrome 3, 603830 Heart block, nonprogressive, 113900 Cardiomyopathy, dilated, 1E, 601154

				Brugada syndrome 1, 601144 Heart block, progressive, type IA, 113900
SDHA	94.1	85.1%	78.0%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011
SGCA	170.3	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	135.1	99.8%	97.9%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	78.2	100.0%	98.2%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	117.7	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHOC2	136.8	100.0%	99.4%	Noonan syndrome-like with loose anagen hair, 607721
SHROOM3	165.5	100.0%	99.3%	No OMIM Disease ID
SKI	149.5	100.0%	99.7%	Shprintzen-Goldberg syndrome, 182212
SLC22A5	144.8	100.0%	100.0%	Carnitine deficiency, systemic primary, 212140
SLC25A20	96.7	100.0%	99.9%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	141.9	100.0%	99.9%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC2A10	167.7	98.0%	97.6%	Arterial tortuosity syndrome, 208050
SLMAP	123.9	98.8%	93.0%	No OMIM Disease ID
SMAD1	154.2	99.9%	99.0%	No OMIM Disease ID
SMAD2	126.9	100.0%	99.5%	No OMIM Disease ID
SMAD3	138.0	100.0%	100.0%	Loeys-Dietz syndrome 3, 613795
SMAD4	109.9	100.0%	99.9%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	209.5	99.9%	95.6%	Aortic valve disease 2, 614823
SMAD9	117.2	100.0%	99.8%	Pulmonary hypertension, primary, 2, 615342
SNTA1	103.0	99.0%	92.1%	Long QT syndrome 12, 612955
SOS1	100.6	99.7%	96.7%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRI	119.2	99.5%	96.7%	No OMIM Disease ID

SYNE1	123.5	98.3%	97.8%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Arthrogryposis multiplex congenita, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE2	112.1	99.6%	97.9%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
TAB2	171.5	100.0%	99.7%	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	125.3	99.3%	96.2%	Barth syndrome, 302060
TBX1	114.2	93.7%	88.3%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TBX20	112.3	100.0%	100.0%	Atrial septal defect 4, 611363
TBX5	145.6	100.0%	100.0%	Holt-Oram syndrome, 142900
TCAP	113.4	100.0%	100.0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TDGF1	127.1	99.8%	95.5%	Forebrain defects, 0
TECRL	71.0	96.0%	87.5%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	186.8	99.7%	97.6%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TGFB2	179.0	100.0%	99.8%	Loeys-Dietz syndrome 4, 614816
TGFB3	149.0	100.0%	100.0%	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBR1	156.6	97.3%	94.3%	Loeys-Dietz syndrome 1, 609192
TGFBR2	169.1	100.0%	100.0%	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
THBS4	123.6	100.0%	99.8%	No OMIM Disease ID
TLL1	128.5	100.0%	99.9%	Atrial septal defect 6, 613087
TMEM43	138.5	99.8%	98.3%	Emery-Dreifuss muscular dystrophy 7, AD, 614302 Arrhythmogenic right ventricular dysplasia 5, 604400
TMPO	126.2	97.5%	93.1%	No OMIM Disease ID
TNNC1	160.8	100.0%	100.0%	Cardiomyopathy, hypertrophic, 13, 613243 Cardiomyopathy, dilated, 1Z, 611879
TNNI3	112.2	99.6%	96.4%	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210
TNNI3K	103.8	100.0%	99.6%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117

TNNT2	114.9	100.0%	100.0%	Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494 Cardiomyopathy, dilated, 1D, 601494
TOR1AIP1	142.0	99.2%	97.1%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TPM1	120.1	100.0%	99.8%	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878
TRDN	78.9	95.2%	83.7%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	104.5	100.0%	100.0%	No OMIM Disease ID
TRPM4	152.9	100.0%	100.0%	Erythrokeratodermia veriabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559
TSFM	123.3	100.0%	99.6%	Combined oxidative phosphorylation deficiency 3, 610505
TTN	165.0	98.6%	98.1%	Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, familial hypertrophic, 9, 613765 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807
TTR	125.9	94.6%	94.6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TXNRD2	121.5	96.7%	95.6%	?Glucocorticoid deficiency 5, 617825
VCL	105.4	100.0%	99.2%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	144.0	100.0%	100.0%	No OMIM Disease ID
XK	88.8	99.9%	99.5%	McLeod syndrome with or without chronic granulomatous disease, 300842
ZBTB17	158.6	100.0%	100.0%	No OMIM Disease ID
ZFPM2	160.7	100.0%	99.9%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	155.4	100.0%	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 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors