HEART GENE PANEL DG 2.16 (298 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
AARS2	122,7	100.0%	99.8%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCC6	109,1	93.6%	92.8%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	142,6	100.0%	99.7%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABL1	155,2	100.0%	99.9%	Congenital heart defects and skeletal malformations syndrome, 617602 Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 0
ACAD8	122,1	100.0%	99.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	124,3	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	115,8	99.8%	98.0%	VLCAD deficiency, 201475
ACSF3	145,8	99.9%	99.1%	Combined malonic and methylmalonic aciduria, 614265
ACTA2	87,3	99.9%	98.6%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTC1	111,1	100.0%	98.9%	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTN2	128,9	100.0%	100.0%	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACVR2B	115,6	99.7%	97.0%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS19	115,7	98.4%	94.6%	No OMIM phenotype
ADCY5	131,8	97.8%	94.7%	Dyskinesia, familial, with facial myokymia, 606703
AGK	108,5	99.5%	95.7%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	146,9	100.0%	99.4%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400

AGPAT2	162,6	99.1%	94.8%	Lipodystrophy, congenital generalized, type 1, 608594
AKAP9	98,4	99.1%	96.7%	?Long QT syndrome-11, 611820
ALDH1A2	105,1	99.9%	98.6%	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	172,8	100.0%	99.7%	Alstrom syndrome, 203800
ALPK3	113,9	99.4%	97.2%	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	139,5	100.0%	100.0%	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKRD1	98,1	99.9%	98.6%	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
ARIH1	115,3	99.9%	99.1%	No OMIM phenotype
ATPAF2	103,5	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	171,4	100.0%	99.9%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	51,1	96.6%	84.1%	Nestor-Guillermo progeria syndrome, 614008
BGN	135,9	100.0%	99.8%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BSCL2	105,2	100.0%	100.0%	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	112,6	99.7%	98.5%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
CACNA1C	141	99.9%	99.1%	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	127,4	98.0%	97.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474

				Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	94,2	99.0%	94.5%	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	130,1	99.9%	99.2%	Brugada syndrome 4, 611876
CALM1	95,8	99.9%	97.8%	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	44	66.7%	61.3%	Long QT syndrome 15, 616249
CALM3	96,8	100.0%	99.4%	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	113,5	100.0%	99.1%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV1	189,3	100.0%	100.0%	?Lipodystrophy, congenital generalized, type 3, 612526 Lipodystrophy, familial partial, type 7, 606721 Pulmonary hypertension, primary, 3, 615343
CAV3	220,8	100.0%	100.0%	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072
CAVIN4	141,8	100.0%	100.0%	No OMIM phenotype
CDH2	112,6	99.8%	98.5%	No OMIM phenotype
CFAP53	131,8	99.1%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	125,8	91.0%	80.1%	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHKB	115,4	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	115	100.0%	100.0%	No OMIM phenotype
CITED2	149,7	99.2%	99.0%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	99,2	99.3%	96.8%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426

				{Multiple system atrophy, susceptibility to}, 146500
COX15	87,7	99.9%	98.3%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
CPT1A	131,5	99.9%	98.4%	CPT deficiency, hepatic, type IA, 255120
CPT2	139,2	98.3%	98.2%	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	98,9	99.8%	95.9%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRYAB	94	99.7%	96.8%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CSRP3	88,6	100.0%	98.3%	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CTF1	30,1	64.4%	43.0%	No OMIM phenotype Cardiomyopathy,dilated (Erdmann (2000) Hum Mutat 16,448)
CTNNA3	131,7	100.0%	99.8%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
DES	125	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DMD	108,2	99.4%	98.0%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DOLK	157,2	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DPM3	200,5	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	122,4	99.9%	98.6%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	123,7	99.6%	97.2%	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSG2	129,5	99.9%	99.2%	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSP	140,6	100.0%	99.6%	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	125,2	100.0%	99.9%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
EEF1A2	188,3	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFEMP2	129,4	100.0%	100.0%	Cutis laxa, autosomal recessive, type IB, 614437
EHMT1	127,7	94.6%	94.2%	Kleefstra syndrome 1, 610253
ELN	103,1	100.0%	98.9%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
EMD	138,2	100.0%	98.9%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMILIN1	112,2	100.0%	97.6%	No OMIM phenotype Connective tissue disease, autosomal dominant (Capuano (2016) Hum Mutat 37, 84)
ENPP1	129,2	97.5%	93.3%	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	136,9	100.0%	99.9%	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
FAH	128,4	100.0%	99.8%	Tyrosinemia, type I, 276700
FBN1	137,1	100.0%	99.8%	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	142,2	100.0%	99.8%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXO32	138,7	100.0%	100.0%	No OMIM phenotype
FGF12	100,4	100.0%	99.9%	Epileptic encephalopathy, early infantile, 47, 617166
FHL1	64,1	98.3%	91.4%	?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	156,6	99.9%	99.1%	No OMIM phenotype Cardiomyopathy,hypertrophic (Friedrich (2014) Basic Res Cardiol 109,451) ?Distal myopathy (Evila (2016) Neuromuscul Disord 26,7)
FHOD3	132,8	99.9%	98.9%	No OMIM phenotype
FKRP	153,3	100.0%	100.0%	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	107,5	99.7%	96.1%	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	142,7	100.0%	99.9%	?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	153,2	100.0%	99.6%	Cardiomyopathy, familial hypertrophic, 26, 0 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLT4	160,3	99.2%	99.1%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FOXC2	122,3	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	5	32.2%	18.7%	No OMIM phenotype
FOXE3	88,3	89.7%	82.5%	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXH1	84,5	100.0%	99.5%	No OMIM phenotype Congenital heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J Cardiol 145,83)

FOXL1	144	100.0%	99.4%	No OMIM phenotype ?Hypoplastic left heart syndrome (Iascone (2012) Clin Genet 81,542)
GAA	160,8	100.0%	99.9%	Glycogen storage disease II, 232300
GATA4	87,6	95.9%	86.7%	?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	74	100.0%	99.2%	Congenital heart defects, multiple types, 5, 617912
GATA6	110,2	98.3%	92.5%	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	136,1	100.0%	100.0%	?Cardiomyopathy, dilated, 2B, 614672
GATB	99,8	100.0%	99.4%	No OMIM phenotype
GATC	141,7	100.0%	100.0%	No OMIM phenotype
GBE1	157,4	99.9%	99.7%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDF1	50,7	97.8%	84.7%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF2	142,4	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GJA5	207,8	100.0%	100.0%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	73,6	99.5%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GMPPB	211,8	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
GNPTAB	148	100.0%	99.3%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GPD1L	128	100.0%	99.9%	Brugada syndrome 2, 611777
HADHA	72,9	96.3%	89.3%	Fatty liver, acute, of pregnancy, 609016

				HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	77,6	96.7%	83.8%	Trifunctional protein deficiency, 609015
HAND1	162,9	100.0%	100.0%	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	85,1	100.0%	99.2%	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	37	66.9%	54.7%	No OMIM phenotype
HCN3	144,5	100.0%	99.5%	No OMIM phenotype
HCN4	96	100.0%	99.7%	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HEY2	162,7	99.4%	96.6%	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
HFE	108	100.0%	98.9%	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	NC	NC	NC	Hemochromatosis, type 2A, 602390
HSPB6	95,2	96.3%	88.2%	No OMIM phenotype
IDUA	148,1	98.9%	94.6%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
ILK	127,5	100.0%	99.8%	No OMIM phenotype Cardiomyopathy, dilated (Knoll (2007) Circulation 116,515) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89, 476)
ITPA	130,2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAG1	133,7	99.2%	97.1%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500

JPH2	123,7	99.5%	95.5%	Cardiomyopathy, hypertrophic, 17, 613873
JUP	124,5	100.0%	99.8%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KCNA5	154,2	100.0%	100.0%	Atrial fibrillation, familial, 7, 612240
KCND2	158,5	100.0%	100.0%	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	162	99.9%	99.2%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	369,2	100.0%	100.0%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	126,7	100.0%	97.9%	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	143,4	100.0%	100.0%	?Brugada syndrome 6, 613119
KCNE4	115	80.5%	80.5%	No OMIM phenotype ?Periodic paralysis (Silva (2004) Arq Bras Endocrinol Metabol 48,196) {Atrial fibrillation, association with} (Zeng (2007) Cardiology 108,97)
KCNE5	130,8	100.0%	98.7%	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	106,9	98.6%	95.1%	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ11	199,7	100.0%	100.0%	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	154,8	100.0%	100.0%	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ5	160,1	100.0%	99.8%	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ8	122,5	100.0%	100.0%	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	161,7	99.8%	98.5%	Pulmonary hypertension, primary, 4, 615344
KCNN3	120,6	100.0%	99.9%	No OMIM phenotype
KCNQ1	135,8	97.9%	95.3%	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	128	100.0%	99.8%	No OMIM phenotype
KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920
KRAS	67,2	99.4%	97.3%	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	130,6	100.0%	99.5%	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMA4	118,2	100.0%	99.7%	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	92,3	97.9%	92.8%	Danon disease, 300257
LDB3	147,1	96.0%	94.7%	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LEFTY2	69,3	99.5%	91.7%	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
LIMS2	118,5	95.7%	93.2%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMNA	104,7	97.7%	91.9%	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	156,1	100.0%	100.0%	No OMIM phenotype Megacystis-microcolon-intestinal hypoperistalsis syndrome (Halim (2017) Proc Natl Acad Sci USA 114)
LOX	158,5	99.9%	99.6%	Aortic aneurysm, familial thoracic 10, 617168
LRRC10	176,8	100.0%	100.0%	No OMIM phenotype Cardiomyopathy,dilated (Qu (2015) Mol Med Rep 12,3718)
LTBP3	147,5	100.0%	99.6%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	143,6	100.0%	99.7%	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MAT2A	92,5	99.5%	95.7%	No OMIM phenotype Thoracic aortic aneurysms (Guo (2015) Am J Hum Genet 96, 170)
MCTP2	120,6	99.5%	97.4%	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	108,5	99.9%	99.6%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MFAP5	106,8	99.6%	94.8%	Aortic aneurysm, familial thoracic 9, 616166
MIB1	127,1	100.0%	99.9%	Left ventricular noncompaction 7, 615092
MLYCD	95,7	99.4%	96.5%	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	94,9	100.0%	98.0%	Heterotaxy, visceral, 7, autosomal, 616749
MYBPC3	141,5	100.0%	98.6%	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYBPHL	82,2	99.7%	96.2%	No OMIM phenotype
MYH11	122,1	100.0%	99.5%	Aortic aneurysm, familial thoracic 4, 132900
MYH6	95,5	99.0%	95.3%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	92,2	99.5%	96.4%	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	119,3	99.6%	96.4%	No OMIM phenotype ?Cardiomyopathy,left ventricular noncompaction (Esposito (2013) Orphanet J Rare Dis 8) ?Hearing loss (Haraksingh (2014) BMC Genomics 15,1155)
MYL2	132,2	99.8%	95.9%	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	98	100.0%	100.0%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	133,2	100.0%	100.0%	?Atrial fibrillation, familial, 18, 617280
MYL7	135,5	100.0%	100.0%	No OMIM phenotype
MYLK	124,3	99.9%	99.5%	Aortic aneurysm, familial thoracic 7, 613780
MYLK2	132,9	100.0%	100.0%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	126,9	99.6%	98.0%	No OMIM phenotype
MYO6	101,5	99.5%	96.4%	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYOM1	122,3	99.9%	98.7%	No OMIM phenotype
MYOT	138,6	100.0%	99.2%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	142,4	100.0%	99.9%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	124,8	99.9%	99.0%	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
NCOA6	124,7	100.0%	99.8%	No OMIM phenotype
NEBL	97,5	98.5%	95.9%	No OMIM phenotype Cardiomyopathy,dilated (Purejav (2010) J Am Coll Cardiol 56,1493)
NEXN	90,2	96.1%	85.9%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NKX2-5	120,8	100.0%	99.9%	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	139,9	100.0%	100.0%	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NNT	124,6	100.0%	98.5%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NODAL	144,8	100.0%	100.0%	Heterotaxy, visceral, 5, 270100
NOS1AP	196,3	100.0%	100.0%	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	141,8	99.8%	98.9%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	123,7	100.0%	99.6%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPA	158,5	100.0%	100.0%	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPPB	221,6	100.0%	100.0%	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	236,6	100.0%	100.0%	Congenital heart defects, multiple types, 4, 615779
NRAS	145,5	100.0%	100.0%	?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	120,3	98.4%	96.9%	?Atrial fibrillation 15, 615770
OBSCN	148,6	99.9%	99.2%	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	99,2	99.3%	95.5%	Propionicacidemia, 606054
PCCB	111,8	99.3%	96.9%	Propionicacidemia, 606054

PDLIM3	143,9	100.0%	99.6%	No OMIM phenotype Cardiomyopathy,dilated (Arola (2007) Mol Genet Metab 90,435 ?Cardiomyopathy, hypertrophic (Bagnall (2010) Int J Cardiol 145,601)
PDLIM5	120,2	92.3%	89.3%	No OMIM phenotype
PEX5	107,9	100.0%	99.2%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGM1	128,8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PHKA1	90,2	97.4%	91.6%	Muscle glycogenosis, 300559
PHYH	74	99.9%	96.9%	Refsum disease, 266500
PITX2	164,8	100.0%	99.5%	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PKD1L1	108,7	100.0%	99.3%	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	91,9	96.7%	90.7%	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	119,2	99.7%	96.9%	No OMIM phenotype
PLD1	116,4	99.9%	99.3%	Cardiac valvular defect, developmental, 212093
PLEKHM2	131,2	100.0%	100.0%	No OMIM phenotype Cardiomyopathy, dilated with left ventricular noncompaction (Muhammad (2015) Hum Mol Genet 24, 7227)
PLN	163,5	100.0%	100.0%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	131,9	99.8%	97.3%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PMEPA1	114,4	100.0%	99.2%	No OMIM phenotype
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	142,7	100.0%	99.8%	Neutral lipid storage disease with myopathy, 610717
POMT1	130,6	99.7%	97.8%	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	103,3	100.0%	98.4%	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	92,3	98.8%	91.7%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	148,5	100.0%	99.3%	Cardiomyopathy, dilated, 2C, 618189

PRDM16	205,4	99.9%	99.1%	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRKAG2	129,9	99.7%	97.4%	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKG1	125,4	99.7%	98.4%	Aortic aneurysm, familial thoracic 8, 615436
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
QRSL1	85,2	98.6%	93.1%	No OMIM phenotype Infantile mitochondrial disorder, lethal (Kohda (2016) PLoS Genet 12, e1005679)
RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RANGRF	142,2	100.0%	99.4%	No OMIM phenotype Brugada syndrome (Selga (2015) PLoS One 10,e0132888 Histiocytoid cardiomyopathy (Cataldo (2014)
RBM20	177,6	100.0%	99.7%	Cardiomyopathy, dilated, 1DD, 613172
RIT1	139,2	100.0%	100.0%	Noonan syndrome 8, 615355
RYR2	124,9	99.9%	98.9%	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
SCN10A	133,3	100.0%	99.4%	Episodic pain syndrome, familial, 2, 615551
SCN1B	169,7	99.9%	98.1%	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	176,4	100.0%	100.0%	Atrial fibrillation, familial, 14, 615378
SCN3B	137,7	100.0%	100.0%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	66	99.8%	97.7%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	141,1	99.0%	99.0%	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	88,9	85.1%	77.7%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SGCA	158,4	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	140,1	99.3%	96.7%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	78	99.8%	97.2%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	114,5	100.0%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHOC2	139,6	99.9%	99.4%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM3	151,4	99.9%	99.1%	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	132,9	100.0%	99.3%	Shprintzen-Goldberg syndrome, 182212
SLC22A5	129,7	100.0%	100.0%	Carnitine deficiency, systemic primary, 212140
SLC25A20	90,7	100.0%	100.0%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	130,9	100.0%	99.9%	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	152,6	98.0%	97.5%	Arterial tortuosity syndrome, 208050
SLMAP	122,5	99.2%	94.8%	No OMIM phenotype Brugada syndrome (Ishikawa (2012) Circ Arrhythm Electrophysiol epub)
SMAD1	147,5	99.9%	98.8%	No OMIM phenotype
SMAD2	127,3	100.0%	99.3%	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	126,7	100.0%	99.8%	Loeys-Dietz syndrome 3, 613795
SMAD4	108,9	100.0%	99.9%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210

				Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	180,5	98.8%	89.1%	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMAD9	110	100.0%	100.0%	Pulmonary hypertension, primary, 2, 615342
SNTA1	94,2	97.5%	89.0%	Long QT syndrome 12, 612955
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SRI	119,4	99.8%	96.2%	No OMIM phenotype
SYNE1	121,6	98.3%	97.8%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	110,7	99.6%	98.0%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
TAB2	170,6	99.9%	99.5%	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	114,5	99.3%	95.8%	Barth syndrome, 302060
TBX1	101,2	93.0%	86.9%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX20	108,2	100.0%	99.9%	Atrial septal defect 4, 611363
TBX5	135,3	100.0%	100.0%	Holt-Oram syndrome, 142900
TCAP	100,1	100.0%	100.0%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TDGF1	120,9	99.7%	94.8%	Forebrain defects, 0
TECRL	75	97.1%	90.3%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	168,3	99.2%	96.8%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TGFB2	173,2	99.9%	99.0%	Loeys-Dietz syndrome 4, 614816
TGFB3	140,4	100.0%	100.0%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	156,4	95.4%	93.8%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	156,8	100.0%	100.0%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TLL1	129,8	100.0%	99.9%	Atrial septal defect 6, 613087
TMEM43	131	99.9%	98.0%	Arrhythmogenic right ventricular dysplasia 5, 604400

				Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMPO	122,4	97.7%	93.9%	?Cardiomyopathy, dilated, 1T, 613740
TNNC1	150,1	100.0%	100.0%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	107,3	99.6%	94.9%	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690
TNNI3K	105,8	99.9%	99.3%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	106,4	100.0%	100.0%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TOR1AIP1	133,3	99.5%	97.2%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TPM1	113	100.0%	99.2%	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TRDN	82,7	97.5%	88.4%	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	99,9	100.0%	100.0%	No OMIM phenotype Hypertrophic cardiomyopathy (Chen (2012) Circ Res 111,907)
TRPM4	137,4	100.0%	99.9%	Progressive familial heart block, type IB, 604559
TSFM	120	100.0%	99.2%	Combined oxidative phosphorylation deficiency 3, 610505
TTN	163	98.6%	98.1%	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334
TTR	122,6	94.6%	94.6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TXNRD2	111,6	96.7%	95.2%	?Glucocorticoid deficiency 5, 617825
VCL	100,5	99.9%	98.9%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	144,4	100.0%	99.9%	No OMIM phenotype ?Schizphrenia (Fromer (2014) Nature 506,179)
XK	85,4	100.0%	99.4%	McLeod syndrome with or without chronic granulomatous disease, 300842

ZBTB17	143	100.0%	100.0%	No OMIM phenotype
ZFPM2	155,6	100.0%	99.8%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	140,9	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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors