HEART GENE PANEL DG 3.1.0 (308 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	100	99,4	100	100	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCC6	93,6	92,4	100	100	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC9	100	99,9	100	100	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABL1	100	100	100	100	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACAD8	100	100	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	99,4	97,3	100	100	VLCAD deficiency, 201475
ACSF3	100	99,9	100	100	Combined malonic and methylmalonic aciduria, 614265
ACTA2	100	99	100	100	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTC1	100	99,7	100	100	Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098
ACTN2	100	100	100	100	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	98,3	95	100	100	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS19	95	91,6	100	100	No OMIM disease ID
ADCY5	95,1	91,2	99,2	98	Dyskinesia, familial, with facial myokymia, 606703

AGK	90,6	88,6	91,2	91,2	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	100	99,4	100	100	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGPAT2	99,6	96,1	100	100	Lipodystrophy, congenital generalized, type 1, 608594
AKAP9	98,8	95,5	100	100	?Long QT syndrome 11, 611820
ALDH1A2	99,9	98,5	100	100	No OMIM disease ID
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
ALPK3	97,8	94,6	100	100	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	100	100	100	100	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919
ANKRD1	100	99,4	100	100	No OMIM disease ID
ARIH1	100	99,5	100	100	No OMIM disease ID
ATPAF2	100	100	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	100	100	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	98,3	86,6	100	100	Nestor-Guillermo progeria syndrome, 614008
BGN	100	100	100	100	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BMPR2	99,9	99,9	99,9	99,9	Pulmonary venoocclusive disease 1, 265450 Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600
BRAF	91	81,1	100	100	Melanoma, malignant, somatic, 155600 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 114500 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0
BSCL2	100	100	100	100	Neuropathy, distal hereditary motor, type VC, 619112 Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BVES	99,9	98,8	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
CACNA1C	99,9	99,2	100	100	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875

CACNA1D	98	97,9	100	100	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	98,6	95,3	100	100	No OMIM disease ID
CACNB2	98,6	98,5	100	100	Brugada syndrome 4, 611876
CALM1	100	99,4	100	100	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	67,8	65,1	72	72	Long QT syndrome 15, 616249
CALM3	100	99,1	100	100	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CASQ2	100	100	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CAV1	100	100	100	100	Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343
CAV3	100	100	100	100	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	99,3	97,7	100	100	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CFAP53	99,6	97,4	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	84,2	74,1	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHKB	100	99,7	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	100	99,9	100	100	No OMIM disease ID
CITED2	99,2	99	100	100	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	99,6	97,6	100	100	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COQ2	98	95,3	97,2	97,2	{Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426
COX15	99,9	98,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CPT1A	100	98,9	100	100	CPT deficiency, hepatic, type IA, 255120
CPT2	98,2	97,8	100	100	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212

CRELD1	99,9	95	100	100	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRYAB	100	99,2	100	100	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CSRP3	100	99,1	100	100	Cardiomyopathy, hypertrophic, 12, 612124 ?Cardiomyopathy, dilated, 1M, 607482
CTNNA3	100	99,8	100	100	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
DCHS1	99,8	99,1	100	100	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DES	100	99,7	100	100	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DMD	99,6	98,6	100	100	Cardiomyopathy, dilated, 3B, 302045 Becker muscular dystrophy, 300376 Duchenne muscular dystrophy, 310200
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DPM3	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992
DPP6	99,7	97,8	99,4	97,6	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	99,8	98,4	100	100	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	100	99,6	100	100	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSP	100	99,6	100	100	Keratosis palmoplantaris striata II, 612908 Arrhythmogenic right ventricular dysplasia 8, 607450 Epidermolysis bullosa, lethal acantholytic, 609638 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Skin fragility-woolly hair syndrome, 607655 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DTNA	100	99,9	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DZIP1	98,4	96,6	100	100	?Mitral valve prolapse 3, 610840 Spermatogenic failure 47, 619102
EEF1A2	100	100	99,9	99,1	Developmental and epileptic encephalopathy 33, 616409 Mental retardation, autosomal dominant 38, 616393

EFEMP2	100	100	100	100	Cutis laxa, autosomal recessive, type IB, 614437
EHMT1	94,5	93,7	99,6	99,5	Kleefstra syndrome 1, 610253
ELN	99,8	97,8	100	100	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
EMD	99,9	98,4	100	99,1	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMILIN1	99,3	89,8	100	100	No OMIM disease ID
ENPP1	96,4	91,2	98,7	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853
FAH	100	100	100	100	Tyrosinemia, type I, 276700
FBN1	100	99,9	100	100	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	100	99,9	100	100	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXO32	100	100	100	100	No OMIM disease ID
FGF12	99,9	98,1	100	100	Developmental and epileptic encephalopathy 47, 617166
FHL1	99,7	95,8	100	100	Scapuloperoneal myopathy, X-linked dominant, 300695 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 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 Myopathy, X-linked, with postural muscle atrophy, 300696
FHL2	99,9	98,7	100	100	No OMIM disease ID
FHOD3	100	99,6	100	100	No OMIM disease ID
FKRP	100	100	100	99,9	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	99,7	97	100	100	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	100	99,9	100	100	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	100	99,6	100	100	Cardiomyopathy, familial hypertrophic, 26, 617047 Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065
FLT4	99,2	98,3	100	100	Congenital heart defects, multiple types, 7, 618780 Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FNIP1	100	99,8	100	100	No OMIM disease ID
FOXC2	100	96,7	100	99,8	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	26,3	13,3	100	100	No OMIM disease ID
FOXE3	82,6	72	94,4	87,8	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXH1	100	96,5	100	100	No OMIM disease ID
FOXL1	96,6	89	100	100	No OMIM disease ID
GAA	100	99,9	100	100	Glycogen storage disease II, 232300
GATA4	84,1	74,5	100	99,9	?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	99,7	93,7	100	100	Congenital heart defects, multiple types, 5, 617912
GATA6	89,8	83	99,6	98	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	99,9	97,9	100	99,1	?Cardiomyopathy, dilated, 2B, 614672
GATB	100	99,7	100	100	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100	100	100	100	Combined oxidative phosphorylation deficiency 42, 618839
GBE1	100	99,6	100	100	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GDF1	73,9	54	98,7	92	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GDF2	100	100	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJA5	100	100	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	91,1	88,2	91,3	91,3	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	99,9	97,4	100	100	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GMPPB	100	100	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
GNB2	100	100	100	100	No OMIM disease ID
GNPTAB	100	99,9	100	100	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GPD1L	100	99,8	100	100	Brugada syndrome 2, 611777
HADHA	97,2	91,6	100	100	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016
HADHB	98,8	89,7	100	100	Trifunctional protein deficiency, 609015

HAND1	100	100	100	100	No OMIM disease ID
HAND2	99,8	92,6	100	100	No OMIM disease ID
HCN2	59,2	49,5	84,1	77,3	No OMIM disease ID
HCN3	99,9	98,5	100	100	No OMIM disease ID
HCN4	100	99,3	100	99,9	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HEY2	100	99,3	100	100	No OMIM disease ID
HFE	100	99,7	100	100	{Porphyria variegata, susceptibility to}, 176200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200
HJV	100	100	100	100	Hemochromatosis, type 2A, 602390
HSPB6	91,1	81	100	100	No OMIM disease ID
IDUA	93,7	86,8	100	100	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
ILK	100	100	100	100	No OMIM disease ID
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647 [Inosine triphosphatase deficiency], 613850
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JPH2	95,5	80,3	100	100	Cardiomyopathy, hypertrophic, 17, 613873
JUP	100	99,5	100	100	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KCNA5	100	98,5	100	100	Atrial fibrillation, familial, 7, 612240
KCND2	100	100	100	100	No OMIM disease ID
KCND3	100	99,4	100	100	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	100	100	100	100	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNE2	100	97,2	100	100	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	100	100	100	100	?Brugada syndrome 6, 613119
KCNE4	80,5	80,4	100	100	No OMIM disease ID

KCNE5	98,6	91,8	100	100	No OMIM disease ID
KCNH2	95,8	91,9	100	100	{Long QT syndrome 2, acquired, susceptibility to}, 613688 Long QT syndrome 2, 613688 Short QT syndrome 1, 609620
KCNJ11	100	100	100	100	Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ2	100	100	100	100	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KCNJ5	100	100	100	100	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ8	100	100	100	100	No OMIM disease ID
KCNK3	97,5	95	100	100	Pulmonary hypertension, primary, 4, 615344
KCNN3	100	99,7	100	100	Zimmermann-Laband syndrome 3, 618658
KCNQ1	93,3	90,6	100	99,8	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500 Atrial fibrillation, familial, 3, 607554
KLF10	100	99,9	100	100	No OMIM disease ID
KLHL24	100	100	100	100	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 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	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA4	100	99,9	100	100	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	99,2	95,6	100	100	Danon disease, 300257
LDB3	95,4	94,7	100	100	Cardiomyopathy, hypertrophic, 24, 601493 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Myopathy, myofibrillar, 4, 609452 Left ventricular noncompaction 3, 601493
LEFTY2	88,9	81,4	100	100	No OMIM disease ID
LIMS2	93	92,7	99,8	98,9	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMNA	97,4	91,9	100	100	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	100	100	100	100	No OMIM disease ID
LOX	100	99,6	100	100	Aortic aneurysm, familial thoracic 10, 617168
LRRC10	100	100	100	100	No OMIM disease ID
LTBP3	99,6	98,1	100	100	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAT2A	99,6	96,4	100	100	No OMIM disease ID
MCTP2	99,7	98,2	100	100	No OMIM disease ID
MED13L	100	99,8	100	100	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MFAP5	99,9	97,6	100	100	Aortic aneurysm, familial thoracic 9, 616166
MIB1	100	99,9	100	100	Left ventricular noncompaction 7, 615092
MLYCD	96	90,4	100	98,9	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	99,9	98,8	100	100	Heterotaxy, visceral, 7, autosomal, 616749

MYBPC3	99,9	97,6	100	100	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYBPHL	99,9	98,6	100	100	No OMIM disease ID
MYH11	100	100	100	100	Aortic aneurysm, familial thoracic 4, 132900
MYH6	99,4	97,1	100	100	Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090 Cardiomyopathy, dilated, 1EE, 613252
MYH7	99,6	97,3	100	100	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	98,4	94,2	100	100	No OMIM disease ID
MYL2	96,1	88,1	99	95,4	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	100	100	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100	100	100	100	?Atrial fibrillation, familial, 18, 617280
MYL7	100	100	100	100	No OMIM disease ID
MYLK	100	99,9	100	100	Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210
MYLK2	100	100	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	99,4	97,8	100	100	No OMIM disease ID
MYO6	99,5	96,6	100	100	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYOM1	99,9	98,4	100	100	No OMIM disease ID
MYOT	100	99,6	100	100	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	100	100	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	100	99,7	100	100	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
MYRF	99,3	98,5	100	100	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113

NAA15	95,8	91	96,8	96,7	Mental retardation, autosomal dominant 50, 617787
NEBL	99,2	97,1	100	100	No OMIM disease ID
NEXN	92	77,5	100	99,9	Cardiomyopathy, hypertrophic, 20, 613876 Cardiomyopathy, dilated, 1CC, 613122
NKX2-5	100	99,7	100	100	Atrial septal defect 7, with or without AV conduction defects, 108900 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
NKX2-6	100	99,5	100	100	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NNT	96,4	95,9	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NODAL	100	100	100	100	Heterotaxy, visceral, 5, 270100
NOS1AP	100	99,9	100	100	Nephrotic syndrome, type 22, 619155
NOTCH1	99,2	97,2	100	100	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	100	99,5	100	100	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPPA	100	100	100	100	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPPB	100	100	100	100	No OMIM disease ID
NR2F2	100	98,5	100	100	Congenital heart defects, multiple types, 4, 615779 46,XX sex reversal 5, 618901
NRAS	100	100	100	100	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	99,2	97,4	100	100	?Atrial fibrillation 15, 615770
OBSCN	99,3	98,1	100	99,9	No OMIM disease ID
PCCA	99,5	96,7	100	100	Propionicacidemia, 606054
PCCB	97,9	96	98,7	96,2	Propionicacidemia, 606054
PDLIM3	100	99,7	100	100	No OMIM disease ID
PDLIM5	93,5	91,3	97,5	95,2	No OMIM disease ID

PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGM1	94,2	94,2	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PHKA1	99,2	95,3	100	99,9	Muscle glycogenosis, 300559
PHYH	100	99,6	100	100	Refsum disease, 266500
PITX2	99,9	97,7	100	100	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PKD1L1	100	99,8	100	100	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	95,4	88,6	95	95	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	99,8	98,2	100	100	No OMIM disease ID
PLD1	100	99,6	100	100	Cardiac valvular defect, developmental, 212093
PLEKHM2	100	100	100	100	No OMIM disease ID
PLN	100	100	100	100	Cardiomyopathy, hypertrophic, 18, 613874 Cardiomyopathy, dilated, 1P, 609909
PLOD1	100	98,4	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PMEPA1	100	99,2	100	99,9	No OMIM disease ID
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	99,7	96,1	100	100	Neutral lipid storage disease with myopathy, 610717
POMT1	99,3	97,5	100	100	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	99,4	96,4	100	100	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	98,7	94	100	100	Sudden cardiac failure, infantile, 617222 ?Sudden cardiac failure, alcohol-induced, 617223
PPCS	99,8	99,5	100	100	Cardiomyopathy, dilated, 2C, 618189
PRDM16	99,8	99,1	100	100	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRKAG2	99,1	96,7	100	99,4	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKG1	92,5	91,2	92,7	92,7	Aortic aneurysm, familial thoracic 8, 615436

PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
QRSL1	99,2	93,9	100	100	Combined oxidative phosphorylation deficiency 40, 618835
RAF1	100	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RANGRF	100	99,9	100	100	No OMIM disease ID
RBM20	100	99,9	100	100	Cardiomyopathy, dilated, 1DD, 613172
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RRAD	85,4	80,7	99,4	96,2	No OMIM disease ID
RRAGC	100	99,7	100	100	No OMIM disease ID
RYR2	99,9	99	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Arrhythmogenic right ventricular dysplasia 2, 600996
SCN10A	100	99,6	100	100	Episodic pain syndrome, familial, 2, 615551
SCN1B	98	96,4	99,8	99,3	Atrial fibrillation, familial, 13, 615377 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2B	100	100	100	100	Atrial fibrillation, familial, 14, 615378
SCN3B	100	100	100	100	Brugada syndrome 7, 613120 Atrial fibrillation, familial, 16, 613120
SCN4B	100	99,6	100	100	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN5A	99	99	100	100	Atrial fibrillation, familial, 10, 614022 Sick sinus syndrome 1, 608567 {Sudden infant death syndrome, susceptibility to}, 272120 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	85,8	80,4	100	100	Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165 Mitochondrial complex II deficiency, nuclear type 1, 252011

SGCA	100	99,9	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	97,7	96,5	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100	98,9	100	100	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100	99,2	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHOC2	99,9	99,4	100	100	Noonan syndrome-like with loose anagen hair 1, 607721
SHROOM3	98,6	97,8	100	100	No OMIM disease ID
SKI	99,3	94,9	100	99,4	Shprintzen-Goldberg syndrome, 182212
SLC22A5	100	100	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A20	100	100	100	100	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	100	100	100	100	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	97,7	97,7	100	100	Arterial tortuosity syndrome, 208050
SLMAP	99,2	94,6	100	100	No OMIM disease ID
SMAD1	100	99,1	100	100	No OMIM disease ID
SMAD2	100	99,6	100	100	No OMIM disease ID
SMAD3	99,9	99	100	100	Loeys-Dietz syndrome 3, 613795
SMAD4	100	99,9	100	100	Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350
SMAD6	90,9	81	100	99,6	Aortic valve disease 2, 614823 {Radioulnar synostosis, nonsyndromic}, 179300 {Craniosynostosis 7, susceptibility to}, 617439
SMAD9	100	99,9	100	100	Pulmonary hypertension, primary, 2, 615342
SNTA1	87	78,8	99,3	97,2	Long QT syndrome 12, 612955
SOD2	100	100	100	100	{Microvascular complications of diabetes 6}, 612634
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRI	99,9	97,8	100	100	No OMIM disease ID
SYNE1	98,2	97,8	98,8	98,8	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE2	99,7	98,1	100	99,9	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
TAB2	100	99,7	100	100	Congenital heart defects, nonsyndromic, 2, 614980
TAZ	99,1	95,5	100	100	Barth syndrome, 302060

TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TBX20	100	99,7	100	100	Atrial septal defect 4, 611363
TBX5	100	100	100	100	Holt-Oram syndrome, 142900
TCAP	100	100	100	100	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TDGF1	99,9	96,7	100	100	Forebrain defects, 0
TECRL	96,3	89,3	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	99,9	98,6	100	100	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TGFB2	100	100	100	100	Loeys-Dietz syndrome 4, 614816
TGFB3	100	100	100	100	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBR1	93,7	93,6	99	96,3	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	100	100	100	100	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
THBS4	99,9	99,4	100	100	No OMIM disease ID
TJP1	100	99,7	100	100	No OMIM disease ID
TLL1	100	100	100	100	Atrial septal defect 6, 613087
TMEM43	99,9	98,9	100	100	Emery-Dreifuss muscular dystrophy 7, AD, 614302 Arrhythmogenic right ventricular dysplasia 5, 604400
TMPO	98,4	94,7	100	100	No OMIM disease ID
TNNC1	100	100	100	100	Cardiomyopathy, hypertrophic, 13, 613243 Cardiomyopathy, dilated, 1Z, 611879
TNNI3	99,7	95,4	100	100	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210
TNNI3K	100	99,4	100	100	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	94,8	91,1	100	99,3	Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494 Cardiomyopathy, dilated, 1D, 601494
TOR1AIP1	99,9	98	100	100	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072

TPM1	100	99,4	100	99,9	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878
TRDN	96,2	86,7	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	100	100	100	100	No OMIM disease ID
TRPM4	100	99,5	100	100	Erythrokeratodermia veriabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559
TSFM	100	99,5	94,9	94,9	Combined oxidative phosphorylation deficiency 3, 610505
TTN	98,6	98,1	100	100	Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705
TTR	94,6	94,6	94,6	94,6	Amyloidosis, hereditary, transthyretin-related, 105210 [Dystransthyretinemic hyperthyroxinemia], 145680 Carpal tunnel syndrome, familial, 115430
TXNRD2	96,8	95,9	100	100	?Glucocorticoid deficiency 5, 617825
VCL	99,9	99	100	100	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	100	99,9	100	99,9	No OMIM disease ID
XK	99,8	98,1	100	100	McLeod syndrome with or without chronic granulomatous disease, 300842
ZBTB17	100	100	100	100	No OMIM disease ID
ZFPM2	100	100	100	100	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	100	99,9	100	100	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors