HEART GENE PANEL DG 3.2.0 (313 genes)

Releasedate: 16-09-2021

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	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABCC6	93,6	92,5	100	100	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	100	99,8	100	100	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050
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	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	99,7	96,6	100	100	VLCAD deficiency, 201475
ACSF3	100	99,5	100	100	Combined malonic and methylmalonic aciduria, 614265
ACTA2	100	98,9	100	100	Multisystemic smooth muscle dysfunction syndrome, 613834 Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042
ACTC1	99,9	98,9	100	100	Left ventricular noncompaction 4, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424
ACTN2	100	100	100	100	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654
ACVR2B	98,6	95,1	100	100	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS19	95,7	92,6	100	100	No OMIM disease ID
ADCY5	95,9	92,5	99,2	97,9	Dyskinesia, familial, with facial myokymia, 606703

AGK	90,4	87,9	91,2	91,1	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	99,8	99,5	100	100	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	99	94	100	100	Lipodystrophy, congenital generalized, type 1, 608594
AKAP9	98,4	94,6	100	100	?Long QT syndrome 11, 611820
ALDH1A2	100	99	100	100	No OMIM disease ID
ALMS1	99,7	99,5	100	100	Alstrom syndrome, 203800
ALPK3	98,1	95,1	100	100	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	100	99,9	100	100	Long QT syndrome 4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANKRD1	99,9	98,3	100	100	No OMIM disease ID
ARIH1	99,7	99,3	100	100	No OMIM disease ID
ATPAF2	100	99,9	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	100	99,7	100	100	Cardiomyopathy, dilated, 1HYPOGONADOTROPIC HYPOGONADISM, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	95,3	78,1	100	100	Nestor-Guillermo progeria syndrome, 614008
BGN	100	99,9	100	100	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BMPR2	99,9	99,9	99,9	99,9	Pulmonary hypertension, familial primary, 1, with or without HYPOGONADOTROPIC HYPOGONADISMT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BRAF	89,4	77,6	100	100	Melanoma, malignant, somatic, 155600 LEOPARD syndrome 3, 613707 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 Noonan syndrome 7, 613706 Colorectal cancer, somatic, 114500 Nonsmall cell lung cancer, somatic, 211980
BSCL2	100	99,9	100	100	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BVES	99,4	98,6	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	97,9	97,7	100	100	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	99	96	100	99,8	No OMIM disease ID
CACNB2	98,5	98,4	100	100	Brugada syndrome 4, 611876
CALM1	99,9	97,3	100	99,9	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 Long QT syndrome 14, 616247
CALM2	66,5	59,7	72	72	Long QT syndrome 15, 616249
CALM3	100	99,2	100	100	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CASQ2	100	99,8	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASZ1	96,5	93,2	97,8	95,9	No OMIM disease ID
CAV1	100	100	100	100	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721
CAV3	100	100	100	100	Myopathy, distal, Tateyama type, 614321 Creatine phosphokinase, elevated serum, 123320 Cardiomyopathy, familial hypertrophic, 192600 Rippling muscle disease 2, 606072 Long QT syndrome 9, 611818
CDH2	99,4	97,1	100	100	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CFAP53	99,3	96,6	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	85	78	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	100	99,2	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHKB	100	99,6	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	100	100	100	100	No OMIM disease ID
CITED2	99,2	99,1	100	100	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	99,6	96,2	100	100	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COQ2	97,6	96,7	97,2	97,2	Coenzyme Q10 deficiency, primary, 1, 607426

COX15	99,9	97,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CPT1A	99,8	97,6	100	100	CPT deficiency, hepatic, type IA, 255120
CPT2	98,2	97,4	100	100	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CRELD1	99,5	94	100	100	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRYAB	100	98,2	100	100	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Cardiomyopathy, dilated, 1II, 615184
CSRP3	98,9	93,9	100	100	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CTNNA3	99,9	99,8	100	100	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
DCHS1	99,9	99,4	100	100	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DES	100	99,6	100	100	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DMD	99,5	98,1	100	99,9	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DPM3	100	100	100	100	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	99,7	97,9	98,8	96,8	Mental retardation, autosomal dominant 33, 616311
DSC2	99,4	97,4	100	99,9	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	99,8	99,6	100	100	Cardiomyopathy, dilated, 1BB, 612877 Arrhythmogenic right ventricular dysplasia 10, 610193
DSP	99,9	99,4	100	100	Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908

					Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
DTNA	99,9	99,9	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DZIP1	98,3	95,9	100	100	Spermatogenic failure 47, 619102 ?Mitral valve prolapse 3, 610840
EEF1A2	100	100	100	99,4	Mental retardation, autosomal dominant 38, 616393 Developmental and epileptic encephalopathy 33, 616409
EFEMP2	100	100	100	100	Cutis laxa, autosomal recessive, type IB, 614437
EHMT1	94,5	93,6	99,6	99,5	Kleefstra syndrome 1, 610253
ELN	99,8	98,3	100	100	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
EMD	99,8	97,9	100	99,8	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMILIN1	98,3	90,3	100	100	No OMIM disease ID
ENPP1	96,5	90,6	98,8	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
FAH	100	99,5	100	99,9	Tyrosinemia, type I, 276700
FBN1	100	99,7	100	100	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900
FBN2	100	99,8	100	100	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FBXO32	100	100	100	100	No OMIM disease ID
FGF12	100	99,1	100	100	Developmental and epileptic encephalopathy 47, 617166
FHL1	99,4	93,8	100	100	Myopathy, X-linked, with postural muscle atrophy, 300696 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717

FHL2	99,8	98,1	100	100	No OMIM disease ID
FHOD3	100	99,4	100	100	Cardiomyopathy, familial hypertrophic, 28, 619402
FKRP	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	99,8	95,2	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
FLNC	100	99,4	100	100	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLT4	99,2	98,9	100	100	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780
FNIP1	99,9	99,9	100	100	No OMIM disease ID
FOXC2	100	98,1	100	99,6	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	22,4	5	100	100	No OMIM disease ID
FOXE3	87,6	79	95,8	89,2	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968
FOXH1	100	98,7	100	100	No OMIM disease ID
FOXL1	97	88,8	100	100	No OMIM disease ID
GAA	100	99,9	100	100	Glycogen storage disease II, 232300

GATA4	87,4	78,5	100	100	Tetralogy of Fallot, 187500 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430 ?Testicular anomalies with or without congenital heart disease, 615542
GATA5	99,9	95,5	100	100	Congenital heart defects, multiple types, 5, 617912
GATA6	91,5	84,5	99,7	98,4	Atrial septal defect 9, 614475 Persistent truncus arteriosus, 217095 Pancreatic agenesis and congenital heart defects, 600001 Atrioventricular septal defect 5, 614474 Tetralogy of Fallot, 187500
GATAD1	100	98,7	99,9	98,4	?Cardiomyopathy, dilated, 2B, 614672
GATB	100	99	100	100	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100	100	100	100	Combined oxidative phosphorylation deficiency 42, 618839
GBE1	99,9	99,7	100	100	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDF1	80,8	59	98,5	92	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
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 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJA5	100	100	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	91	85,9	91,3	91,3	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLB1	99,2	92,8	100	100	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600

GMPPB	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GNB2	100	100	100	100	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 ?Sick sinus syndrome 4, 619464
GNPTAB	99,9	99,7	100	100	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GPD1L	100	98,8	100	100	Brugada syndrome 2, 611777
HADHA	95,5	88,3	100	100	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	97,7	87	100	99,9	Trifunctional protein deficiency, 609015
HAND1	100	100	100	100	No OMIM disease ID
HAND2	99,7	94,9	100	100	No OMIM disease ID
HCN2	59,8	47,7	84	76,9	Febrile seizures, familial, 2, 602477 Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	99,9	98,5	100	100	No OMIM disease ID
HCN4	100	99,2	100	100	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HEY2	99,8	98,7	100	100	No OMIM disease ID
HFE	99,9	97,8	100	100	Hemochromatosis, 235200
HJV	100	100	100	100	Hemochromatosis, type 2A, 602390
HSPB6	92,7	83	100	100	No OMIM disease ID
IDUA	94,6	87,4	100	100	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
ILK	100	99,9	100	100	No OMIM disease ID
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647
JAG1	97,8	96,7	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JPH2	96	85,6	100	100	Cardiomyopathy, dilated, 2E, 619492 Cardiomyopathy, hypertrophic, 17, 613873

JUP	100	99,8	100	100	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528
KCNA5	100	99,3	100	100	Atrial fibrillation, familial, 7, 612240
KCND2	100	100	100	100	No OMIM disease ID
KCND3	100	98,6	100	100	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399
KCNE1	100	100	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	100	96,1	100	100	Long QT syndrome 6, 613693 Atrial fibrillation, familial, 4, 611493
KCNE3	100	100	100	100	?Brugada syndrome 6, 613119
KCNE4	80,5	80,5	100	100	No OMIM disease ID
KCNE5	98,5	93,5	100	100	No OMIM disease ID
KCNH2	95,9	92,1	100	100	Short QT syndrome 1, 609620 Long QT syndrome 2, 613688
KCNJ11	100	100	100	100	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Maturity-onset diabetes of the young, type 13, 616329 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ2	100	100	100	100	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
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	97,9	95,1	100	100	Pulmonary hypertension, primary, 4, 615344
KCNN3	100	99,7	100	100	Zimmermann-Laband syndrome 3, 618658
KCNQ1	93,5	90,6	99,9	99,4	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
KDR	100	99,7	100	100	Hemangioma, capillary infantile, somatic, 602089
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	99,9	99	100	100	Kabuki syndrome 1, 147920
KRAS	99	97,8	100	100	Gastric cancer, somatic, 137215 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
LAMA2	99,9	99,1	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA4	100	99,7	100	100	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	99,3	96	100	99,7	Danon disease, 300257
LDB3	95,4	94,8	100	100	Left ventricular noncompaction 3, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LEFTY2	94,3	84,3	100	100	No OMIM disease ID
LIMS2	94,1	92,7	100	99,7	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMNA	96,1	90,6	100	100	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Restrictive dermopathy, lethal, 275210 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LMOD1	100	99,8	100	100	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LOX	99,8	99,6	100	100	Aortic aneurysm, familial thoracic 10, 617168

LRRC10	100	100	100	100	No OMIM disease ID
LTBP3	99,8	98,6	100	99,9	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	100	99,9	100	100	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAT2A	99,2	93,8	100	100	No OMIM disease ID
MCTP2	99,4	97,7	100	100	No OMIM disease ID
MED13L	100	99,5	100	100	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MFAP5	100	98,7	100	100	Aortic aneurysm, familial thoracic 9, 616166
MIB1	100	99,6	100	99,9	Left ventricular noncompaction 7, 615092
MLYCD	96,8	92,5	100	99,4	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	99,8	99,2	100	100	Heterotaxy, visceral, 7, autosomal, 616749
MYBPC3	99,8	97,6	100	100	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYBPHL	99,5	96,6	100	100	No OMIM disease ID
MYH11	100	99,7	100	100	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 Aortic aneurysm, familial thoracic 4, 132900 Visceral myopathy 2, 619350
MYH6	99,2	96,1	100	100	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251
MYH7	99,1	96,7	100	100	Laing distal myopathy, 160500 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160
MYH7B	98,3	94,8	100	100	No OMIM disease ID
MYL2	94,8	81,1	99,6	97,3	Cardiomyopathy, hypertrophic, 10, 608758 Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	100	100	100	100	Cardiomyopathy, hypertrophic, 8, 608751

MYL4	100	100	100	100	?Atrial fibrillation, familial, 18, 617280
MYL7	100	99,6	100	100	No OMIM disease ID
MYLK	100	99,6	100	100	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 Aortic aneurysm, familial thoracic 7, 613780
MYLK2	100	100	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	99,7	98,1	100	100	No OMIM disease ID
MYO6	99,1	96,3	100	100	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821
MYOM1	99,7	98	100	100	No OMIM disease ID
MYOT	100	99,2	100	100	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	100	100	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	100	99,5	100	100	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYRF	99	97,8	100	100	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280
NAA15	94,8	91,2	96,8	96,7	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NEBL	99,2	97,1	100	100	No OMIM disease ID
NEXN	87,9	71,5	100	99,8	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NKX2-5	100	100	100	100	Hypoplastic left heart syndrome 2, 614435 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Conotruncal heart malformations, variable, 217095 Ventricular septal defect 3, 614432 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	100	100	100	100	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NNT	96,4	96	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NODAL	100	100	100	100	Heterotaxy, visceral, 5, 270100
NOS1AP	100	100	100	100	Nephrotic syndrome, type 22, 619155

NOTCH1	99,3	97,9	100	100	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	100	99,2	100	100	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
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	99,1	100	100	46,XX sex reversal 5, 618901 Congenital heart defects, multiple types, 4, 615779
NRAS	100	100	100	100	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Melanocytic nevus syndrome, congenital, somatic, 137550 Epidermal nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500
NUP155	98,7	96,2	100	99,9	?Atrial fibrillation 15, 615770
OBSCN	99,4	98,5	100	100	No OMIM disease ID
CCDC114	100	99,8	100	100	Ciliary dyskinesia, primary, 20, 615067
PCCA	98,9	93,4	100	100	Propionicacidemia, 606054
PCCB	96,7	95,4	99	96,2	Propionicacidemia, 606054
PDLIM3	100	99,7	100	100	No OMIM disease ID
PDLIM5	92,9	90,1	96,7	94,4	No OMIM disease ID
PEX5	99,9	98,8	100	100	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	88	81	91,3	91,2	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGM1	94,2	94,1	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PHKA1	97,8	93,4	100	99,6	Muscle glycogenosis, 300559
PHYH	100	98,9	100	100	Refsum disease, 266500

PITX2	99,8	97,2	100	100	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PKD1L1	100	99,3	100	100	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	94,3	86,9	95	95	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	99,2	96,1	100	100	No OMIM disease ID
PLD1	99,8	98,7	100	100	Cardiac valvular defect, developmental, 212093
PLEKHM2	100	99,9	100	100	No OMIM disease ID
PLN	100	100	100	100	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	100	98,2	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PMEPA1	100	98,8	100	99,2	No OMIM disease ID
PMM2	99,8	99,8	100	100	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	99,8	96,1	100	100	Neutral lipid storage disease with myopathy, 610717
POMT1	99,5	97,3	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	99,8	97,3	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150
PPA2	97,3	88,6	100	100	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	100	99,1	100	100	Cardiomyopathy, dilated, 2C, 618189
PRDM16	99,9	99,2	100	100	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRKAG2	99,4	96,1	99,9	99,3	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKG1	92,4	91	92,7	92,7	Aortic aneurysm, familial thoracic 8, 615436
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
QRSL1	98,6	92,8	100	99,9	Combined oxidative phosphorylation deficiency 40, 618835

RAF1	99,9	99,2	100	100	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RANGRF	100	99,2	100	100	No OMIM disease ID
RBM20	100	99,4	100	100	Cardiomyopathy, dilated, 1DD, 613172
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RRAD	88	82,4	99,6	96,7	No OMIM disease ID
RRAGC	99,9	99,4	100	100	No OMIM disease ID
RYR2	99,8	98,8	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Ventricular arrythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 Arrhythmogenic right ventricular dysplasia 2, 600996
SCN10A	99,9	98,5	100	100	Episodic pain syndrome, familial, 2, 615551
SCN1B	98,2	96,3	99,7	98,9	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838
SCN2B	100	100	100	100	Atrial fibrillation, familial, 14, 615378
SCN3B	100	100	100	100	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	99,9	97,1	100	100	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN5A	99	98,7	100	100	Ventricular fibrillation, familial, 1, 603829 Heart block, progressive, type IA, 113900 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Long QT syndrome 3, 603830 Sick sinus syndrome 1, 608567 Brugada syndrome 1, 601144 Atrial fibrillation, familial, 10, 614022
SDHA	84,5	77,9	100	100	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011 Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165
SGCA	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099

SGCB	97,8	96,5	100	99,9	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	99,6	96,5	100	100	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100	99,4	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHOC2	99,8	99,6	100	99,9	Noonan syndrome-like with loose anagen hair 1, 607721
SHROOM3	98,5	97,5	100	100	No OMIM disease ID
SKI	99,7	97,1	100	99,7	Shprintzen-Goldberg syndrome, 182212
SLC22A5	100	99,6	100	100	Carnitine deficiency, systemic primary, 212140
SLC25A20	100	98,9	100	100	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	100	99,8	100	100	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184
SLC2A10	97,7	97,7	100	100	Arterial tortuosity syndrome, 208050
SLC30A5	99,4	96,8	100	99,9	No OMIM disease ID
SLMAP	98,4	92,5	100	99,9	No OMIM disease ID
SMAD1	99,8	98,2	100	100	No OMIM disease ID
SMAD2	100	99,8	100	100	No OMIM disease ID
SMAD3	99,9	98,4	100	100	Loeys-Dietz syndrome 3, 613795
SMAD4	99,9	99,9	100	100	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	90,7	79,3	100	99,9	Aortic valve disease 2, 614823
SMAD9	100	99,3	100	100	Pulmonary hypertension, primary, 2, 615342
SNTA1	92,6	80,2	99,9	98,6	Long QT syndrome 12, 612955
SOD2	100	100	100	100	No OMIM disease ID
SOS1	99,6	97,9	100	99,9	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRI	99,8	96,4	100	100	No OMIM disease ID
SYNE1	98,1	97,5	98,8	98,8	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743

SYNE2	99,4	97,2	100	99,9	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
TAB2	99,8	99,2	100	100	Congenital heart defects, nonsyndromic, 2, 614980
TAF1	99,2	95,7	100	100	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TAZ	99,3	93,7	100	100	Barth syndrome, 302060
TBX1	87,4	77,6	93,7	90,2	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX20	100	99,8	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	98,8	91,8	100	100	Forebrain defects,
TECRL	97,5	91,7	100	99,4	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	98,8	96,4	100	100	Patent ductus arteriosus 2, 617035 Char syndrome, 169100
TGFB2	100	100	100	100	Loeys-Dietz syndrome 4, 614816
TGFB3	100	100	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	93,6	93,6	98,8	97,6	Loeys-Dietz syndrome 1, 609192
TGFBR2	100	99,9	100	100	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
THBS4	100	99,4	100	100	No OMIM disease ID
TJP1	100	99,7	100	100	No OMIM disease ID
TLL1	99,9	99,7	100	100	Atrial septal defect 6, 613087
TMEM43	99,9	98,4	100	100	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMPO	98	93,8	100	100	No OMIM disease ID
TNNC1	100	100	100	100	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	99,6	95,5	100	100	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, hypertrophic, 7, 613690

					Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	99,8	99,6	100	100	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	94,6	90,7	100	99,7	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, hypertrophic, 2, 115195 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TOR1AIP1	99,2	96,1	100	100	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TPM1	100	99,3	100	99,8	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878
TRDN	97,7	89,1	100	99,5	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TRIM63	100	99,9	100	100	No OMIM disease ID
TRPM4	100	99,8	100	100	Progressive familial heart block, type IB, 604559 Erythrokeratodermia veriabilis et progressiva 6, 618531
TSFM	100	99,3	94,9	94,9	Combined oxidative phosphorylation deficiency 3, 610505
TTN	98,5	98	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, familial hypertrophic, 9, 613765 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, dilated, 1G, 604145 Myopathy, myofibrillar, 9, with early respiratory failure, 603689
TTR	94,6	94,6	94,6	94,6	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TXNRD2	96,8	95,9	100	100	?Glucocorticoid deficiency 5, 617825
VCL	99,9	98,5	100	100	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	99,9	99,7	100	99,9	No OMIM disease ID
XK	99,7	97,6	100	100	McLeod syndrome with or without chronic granulomatous disease, 300842
ZBTB17	100	100	100	100	No OMIM disease ID
ZFPM2	100	99,9	100	100	Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067 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.
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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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors