HEART GENE PANEL DG 3.5.0 (317 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	100%	100%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABCC6	100%	100%	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	100%	100%	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 ?Atrial fibrillation, familial, 12, 614050 Intellectual disability and myopathy syndrome, 619719
ABL1	100%	100%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACAD8	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	100%	100%	VLCAD deficiency, 201475
ACSF3	100%	100%	Combined malonic and methylmalonic aciduria, 614265
ACTC1	100%	100%	Left ventricular noncompaction 4, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424
ACTN2	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	100%	100%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS19	100%	100%	Cardiac valvular dysplasia 2, 620067
ADCY5	100%	100%	Dyskinesia with orofacial involvement, autosomal dominant, 606703 Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADNP	100%	100%	Helsmoortel-van der Aa syndrome, 615873

AGK	92%	92%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGPAT2	100%	100%	Lipodystrophy, congenital generalized, type 1, 608594
AKAP9	100%	100%	?Long QT syndrome 11, 611820
ALDH1A2	100%	100%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ALMS1	100%	100%	Alstrom syndrome, 203800
ALPK3	100%	100%	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	100%	100%	Long QT syndrome 4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANKRD1	100%	100%	No OMIM disease ID
ANKRD11	100%	100%	KBG syndrome, 148050
ATPAF2	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	100%	100%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	100%	100%	Nestor-Guillermo progeria syndrome, 614008
BICD2	100%	100%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BMPR2	100%	100%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BRAF	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%	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	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
CACNA1C	100%	100%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 Brugada syndrome 3, 611875

CACNA1D	100%	100%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	100%	100%	No OMIM disease ID
CACNB2	100%	100%	Brugada syndrome 4, 611876
CALM1	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 Long QT syndrome 14, 616247
CALM2	74%	74%	Long QT syndrome 15, 616249
CALM3	100%	100%	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CASQ2	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASZ1	100%	99%	No OMIM disease ID
CAV1	100%	100%	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721
CAV3	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	100%	100%	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 ?Attention deficit-hyperactivity disorder 8, 619957 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CFAP53	100%	100%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	100%	100%	Heterotaxy, visceral, 2, autosomal, 605376
CHD4	100%	100%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHKB	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	99%	98%	No OMIM disease ID
CITED2	100%	100%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
COL3A1	100%	100%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COQ2	96%	96%	Coenzyme Q10 deficiency, primary, 1, 607426
COX15	100%	100%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CPT1A	100%	100%	CPT deficiency, hepatic, type IA, 255120

CPT2	100%	100%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CRELD1	100%	100%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRYAB	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	100%	100%	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CTNNA3	100%	100%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
DCHS1	100%	100%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DES	100%	100%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DMD	100%	99%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DNAJC19	100%	100%	3-methylglutaconic aciduria, type V, 610198
DOLK	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DPM3	100%	100%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	100%	100%	Intellectual developmental disorder, autosomal dominant 33, 616311
DSC2	100%	100%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	100%	100%	Cardiomyopathy, dilated, 1BB, 612877 Arrhythmogenic right ventricular dysplasia 10, 610193
DSP	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	100%	100%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DYRK1A	100%	100%	Intellectual developmental disorder, autosomal dominant 7, 614104
DZIP1	100%	100%	Spermatogenic failure 47, 619102 ?Mitral valve prolapse 3, 610840

EEF1A2	100%	100%	Developmental and epileptic encephalopathy 33, 616409 Intellectual developmental disorder, autosomal dominant 38, 616393
EHMT1	100%	100%	Kleefstra syndrome 1, 610253
ELN	100%	100%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
EMD	100%	100%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
ENPP1	100%	100%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
FAH	100%	100%	Tyrosinemia, type I, 276700
FBN1	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%	100%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FBXO32	100%	100%	No OMIM disease ID
FGF12	100%	100%	Developmental and epileptic encephalopathy 47, 617166
FHL1	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	100%	100%	No OMIM disease ID
FHOD3	100%	100%	Cardiomyopathy, familial hypertrophic, 28, 619402
FKRP	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), 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	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152

FLNA	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%	100%	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLT4	100%	100%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780
FNIP1	100%	100%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXC2	100%	100%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	100%	100%	No OMIM disease ID
FOXH1	100%	100%	No OMIM disease ID
FOXL1	100%	100%	No OMIM disease ID
GAA	100%	100%	Glycogen storage disease II, 232300
GATA4	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	100%	100%	Congenital heart defects, multiple types, 5, 617912
GATA6	100%	100%	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%	100%	?Cardiomyopathy, dilated, 2B, 614672
GATB	100%	100%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100%	100%	Combined oxidative phosphorylation deficiency 42, 618839

GBE1	100%	100%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDF1	100%	100%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF2	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GJA5	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	91%	91%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLB1	100%	100%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLIS1	100%	100%	No OMIM disease ID
GLYR1	100%	100%	No OMIM disease ID
GMPPB	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GNB2	100%	100%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 ?Sick sinus syndrome 4, 619464
GNPTAB	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GPD1L	100%	100%	Brugada syndrome 2, 611777
HADHA	100%	100%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	100%	100%	Trifunctional protein deficiency, 609015
HAND1	100%	100%	No OMIM disease ID
HAND2	100%	100%	No OMIM disease ID
HCN2	94%	92%	Febrile seizures, familial, 2, 602477 Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100%	100%	No OMIM disease ID
HCN4	100%	100%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HEY2	100%	100%	No OMIM disease ID
HFE	100%	100%	Hemochromatosis, 235200

HJV	100%	100%	Hemochromatosis, type 2A, 602390
HSPB6	100%	100%	No OMIM disease ID
HSPD1	100%	100%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
IDUA	100%	100%	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
ILK	100%	100%	No OMIM disease ID
ITPA	100%	100%	Developmental and epileptic encephalopathy 35, 616647
JAG1	100%	100%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JPH2	100%	100%	Cardiomyopathy, dilated, 2E, 619492 Cardiomyopathy, hypertrophic, 17, 613873
JUP	100%	100%	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528
KCNA5	100%	100%	Atrial fibrillation, familial, 7, 612240
KCND2	100%	99%	No OMIM disease ID
KCND3	100%	100%	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399
KCNE1	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	100%	100%	Long QT syndrome 6, 613693 Atrial fibrillation, familial, 4, 611493
KCNE3	100%	100%	?Brugada syndrome 6, 613119
KCNE4	100%	100%	No OMIM disease ID
KCNE5	100%	100%	No OMIM disease ID
KCNH2	100%	100%	Short QT syndrome 1, 609620 Long QT syndrome 2, 613688
KCNJ11	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%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622

KCNJ5	100%	100%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ8	100%	100%	No OMIM disease ID
KCNK3	100%	100%	Pulmonary hypertension, primary, 4, 615344
KCNN3	100%	100%	Zimmermann-Laband syndrome 3, 618658
KCNQ1	100%	100%	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
KDR	100%	100%	Hemangioma, capillary infantile, somatic, 602089
KLHL24	100%	100%	Epidermolysis bullosa simplex 6, generalized, with scarring and hair loss, 617294
KMT2A	100%	100%	Wiedemann-Steiner syndrome, 605130
KMT2D	100%	100%	Kabuki syndrome 1, 147920
KRAS	100%	100%	Gastric cancer, somatic, 613659 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	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA4	100%	100%	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	100%	100%	Danon disease, 300257
LDB3	100%	100%	Left ventricular noncompaction 3, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LEFTY2	100%	100%	No OMIM disease ID
LIMS2	100%	100%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMCD1	100%	100%	No OMIM disease ID
LMNA	100%	100%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140

			Cardiomyopathy, dilated, 1A, 115200 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Restrictive dermopathy 2, 619793 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
LMOD2	100%	100%	Cardiomyopathy, dilated, 2G, 619897
LRRC10	100%	100%	No OMIM disease ID
LZTR1	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MED13L	100%	100%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MIB1	100%	100%	Left ventricular noncompaction 7, 615092
MIPEP	100%	100%	Combined oxidative phosphorylation deficiency 31, 617228
MLYCD	100%	100%	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	100%	100%	Heterotaxy, visceral, 7, autosomal, 616749
MTO1	94%	91%	Combined oxidative phosphorylation deficiency 10, 614702
MUC16	100%	100%	No OMIM disease ID
MYBPC3	100%	100%	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYBPHL	100%	100%	No OMIM disease ID
MYH11	100%	100%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 Aortic aneurysm, familial thoracic 4, 132900 Visceral myopathy 2, 619350
MYH6	100%	100%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251
MYH7	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	100%	100%	No OMIM disease ID

MYL2	100%	100%	Cardiomyopathy, hypertrophic, 10, 608758 Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	100%	100%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100%	100%	?Atrial fibrillation, familial, 18, 617280
MYL7	100%	100%	No OMIM disease ID
MYLK3	100%	100%	No OMIM disease ID
MYO6	100%	100%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821
MYOM1	100%	100%	No OMIM disease ID
MYOT	100%	100%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	100%	100%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	100%	100%	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYRF	100%	100%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280
NAA15	97%	97%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NDUFB11	100%	98%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NEBL	100%	99%	No OMIM disease ID
NEXN	100%	100%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	100%	100%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NKX2-5	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%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095

NODAL	100%	100%	Heterotaxy, visceral, 5, 270100
NOS1AP	100%	100%	Nephrotic syndrome, type 22, 619155
NOTCH1	100%	100%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	100%	100%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPA	100%	100%	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPPB	100%	100%	No OMIM disease ID
NR2F2	100%	100%	46,XX sex reversal 5, 618901 Congenital heart defects, multiple types, 4, 615779
NRAP	100%	100%	No OMIM disease ID
NRAS	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
NSD1	100%	100%	Sotos syndrome, 117550
NUP155	100%	100%	?Atrial fibrillation 15, 615770
OBSCN	100%	100%	No OMIM disease ID
CCDC114	100%	100%	Ciliary dyskinesia, primary, 20, 615067
PCCA	100%	100%	Propionicacidemia, 606054
PCCB	100%	98%	Propionicacidemia, 606054
PDLIM3	100%	100%	No OMIM disease ID
PDLIM5	100%	98%	No OMIM disease ID
PEX5	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	91%	91%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGM1	94%	94%	Congenital disorder of glycosylation, type It, 614921
PHKA1	100%	100%	Muscle glycogenosis, 300559
PHYH	100%	100%	Refsum disease, 266500

PITX2	100%	100%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PKD1L1	100%	100%	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	100%	99%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLD1	100%	100%	Cardiac valvular dysplasia 1, 212093
PLEKHM2	100%	100%	No OMIM disease ID
PLN	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PMM2	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	100%	100%	Neutral lipid storage disease with myopathy, 610717
POMT1	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 impaired intellectual development), type B, 1, 613155
POMT2	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 impaired intellectual development), type B, 2, 613156
PPA2	100%	100%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	100%	100%	Cardiomyopathy, dilated, 2C, 618189
PRDM16	100%	100%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM6	100%	100%	Patent ductus arteriosus 3, 617039
PRKAG2	100%	100%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKD1	100%	100%	Congenital heart defects and ectodermal dysplasia, 617364
PTPN11	100%	100%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
QRSL1	100%	100%	Combined oxidative phosphorylation deficiency 40, 618835
RAF1	100%	100%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RANGRF	100%	100%	No OMIM disease ID
RBFOX2	100%	100%	No OMIM disease ID

RBM20	100%	100%	Cardiomyopathy, dilated, 1DD, 613172
RIT1	100%	100%	Noonan syndrome 8, 615355
RPL3L	100%	100%	Cardiomyopathy, dilated, 2D, 619371
RPS6KB1	100%	100%	No OMIM disease ID
RRAD	100%	100%	No OMIM disease ID
RRAGC	100%	100%	No OMIM disease ID
RYR2	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 Arrhythmogenic right ventricular dysplasia 2, 600996
SCN10A	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN1B	100%	100%	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%	Atrial fibrillation, familial, 14, 615378
SCN3B	100%	100%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4B	100%	100%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819
SCN5A	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
SCO2	100%	100%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SDHA	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%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100%	100%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287

SGCG	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHMT2	100%	100%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC2	100%	100%	Noonan syndrome-like with loose anagen hair 1, 607721
SHROOM3	100%	100%	No OMIM disease ID
SLC22A5	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC25A20	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	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
SLC30A5	100%	100%	No OMIM disease ID
SLC4A3	100%	100%	No OMIM disease ID
SLC6A6	100%	100%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLMAP	100%	100%	No OMIM disease ID
SMAD1	100%	100%	No OMIM disease ID
SMAD6	100%	100%	Aortic valve disease 2, 614823
SMAD9	100%	100%	Pulmonary hypertension, primary, 2, 615342
SMARCA4	100%	100%	Coffin-Siris syndrome 4, 614609
SNTA1	100%	100%	Long QT syndrome 12, 612955
SOD2	100%	100%	No OMIM disease ID
SOS1	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRF	100%	100%	No OMIM disease ID
SRI	100%	100%	No OMIM disease ID
SURF1	100%	100%	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
TAB2	100%	100%	Congenital heart defects, nonsyndromic, 2, 614980
TAF1	100%	100%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TAF1A	100%	100%	No OMIM disease ID
TAZ	100%	100%	Barth syndrome, 302060
TBX1	98%	96%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX20	100%	100%	Atrial septal defect 4, 611363
TBX5	100%	100%	Holt-Oram syndrome, 142900

TCAP	100%	100%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TDGF1	100%	100%	Forebrain defects,
TECRL	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	100%	100%	Patent ductus arteriosus 2, 617035 Char syndrome, 169100
TGFB3	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
THBS4	100%	100%	No OMIM disease ID
TJP1	100%	100%	No OMIM disease ID
TLL1	99%	99%	Atrial septal defect 6, 613087
TMEM260	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TMEM43	100%	100%	Arrhythmogenic right ventricular dysplasia 5, 604400 Auditory neuropathy, autosomal dominant 3, 619832 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMPO	100%	100%	No OMIM disease ID
TNNC1	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	100%	100%	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	100%	100%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	100%	100%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, hypertrophic, 2, 115195 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494
TNS1	100%	100%	No OMIM disease ID
TOR1AIP1	100%	100%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TPM1	100%	100%	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878
TRDN	100%	100%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TRIM63	100%	100%	No OMIM disease ID
TRPM4	100%	100%	Progressive familial heart block, type IB, 604559 Erythrokeratodermia variabilis et progressiva 6, 618531

TSC1	100%	100%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSFM	94%	94%	Combined oxidative phosphorylation deficiency 3, 610505
TTN	100%	99%	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	91%	91%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TXNRD2	100%	100%	?Glucocorticoid deficiency 5, 617825
VCL	100%	100%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
XIRP2	100%	100%	No OMIM disease ID
XK	100%	100%	McLeod syndrome with or without chronic granulomatous disease, 300842
ZBTB17	100%	100%	No OMIM disease ID
ZFPM2	100%	100%	Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500
ZIC3	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.
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.
TWIST X2 is the chemistry used for 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 : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors