HEART DISORDERS PANEL[1] DG-4.1.0 (348 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	100%	100%	100%	99.3%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABCC6	98.4%	98.4%	100%	99.2%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	96%	96%	100%	99.6%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719

ABL1	100%	100%	100%	98.8%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACAD8	100%	100%	100%	98.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	100%	100%	100%	98.3%	VLCAD deficiency, 201475
ACSF3	100%	100%	100%	99.4%	Combined malonic and methylmalonic aciduria, 614265
ACTA1	100%	100%	100%	98.5%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800

ACTA2	100%	100%	100%	99.5%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042
ACTC1	100%	100%	100%	99.4%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACTN2	100%	100%	99.9%	97.4%	Myopathy, distal, 6, adult onset, 618655;Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158;Congenital myopathy 8, 618654;Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158
ACVR2B	100%	100%	100%	99.1%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS19	100%	100%	100%	99.7%	Cardiac valvular dysplasia 2, 620067

ADCY5	97.4%	97.1%	100%	98.2%	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%	100%	99.6%	Helsmoortel-van der Aa syndrome, 615873
AGK	91.7%	91.7%	100%	99.7%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGL	100%	100%	100%	99.7%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400
AGPAT2	100%	100%	100%	98%	Lipodystrophy, congenital generalized, type 1, 608594
AKAP9	100%	100%	100%	99.5%	?Long QT syndrome 11, 611820
ALDH1A2	100%	100%	100%	99.6%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ALMS1	100%	100%	100%	99.7%	Alstrom syndrome, 203800
ALPK3	100%	100%	100%	98.9%	Cardiomyopathy, familial hypertrophic 27, 618052

ANK2	100%	100%	100%	99.6%	Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919
ANKRD1	100%	100%	100%	97.6%	
ANKRD11	100%	100%	100%	98.7%	KBG syndrome, 148050
ATPAF2	100%	100%	100%	98.9%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954
BANF1	100%	100%	100%	97.6%	Nestor-Guillermo progeria syndrome, 614008
BICD2	100%	100%	100%	99.4%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BMPR2	100%	100%	100%	99.7%	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%	99.1%	96.2%	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%	100%	98.9%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
BVES	100%	100%	100%	99.5%	
CACNA1C	100%	100%	100%	99.3%	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%	100%	99.4%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 110, 620149
CACNB2	100%	100%	100%	99%	Brugada syndrome 4, 611876
CALM1	100%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100%	98.8%	Long QT syndrome 15, 616249
CALM3	100%	100%	100%	99.4%	Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CASQ2	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASZ1	99.6%	97.3%	100%	96.7%	

CAV1	74.7%	74.6%	100%	99%	Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721
CAV3	100%	100%	100%	99.3%	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%	100%	99.4%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CFAP45	100%	100%	100%	99.5%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP52	100%	100%	100%	99.7%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	100%	100%	100%	99.6%	Heterotaxy, visceral, 6, autosomal recessive, 614779

CFC1	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376
CHD4	100%	100%	100%	99.3%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHKB	100%	100%	100%	98.9%	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	100%	100%	100%	99.6%	
CIROP	98.4%	93.9%	100%	99.5%	Heterotaxy, visceral, 12, autosomal, 619702
CITED2	100%	100%	100%	98.8%	Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431
COL3A1	100%	100%	100%	99.2%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
COQ2	96.3%	96.3%	100%	98.7%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426

COQ7	100%	100%	100%	98.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COX15	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CPT1A	100%	100%	100%	99.3%	CPT deficiency, hepatic, type IA, 255120
CPT2	100%	100%	100%	99.6%	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110
CRELD1	100%	100%	100%	99%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217

CRYAB	100%	100%	100%	99.7%	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%	100%	99.7%	?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124
CTNNA3	100%	100%	100%	99.8%	Arrhythmogenic right ventricular dysplasia 13, 615616
CTNND1	100%	100%	100%	99.6%	Blepharocheilodontic syndrome 2, 617681
DAND5	87.8%	87.8%	100%	99.4%	
DCHS1	100%	100%	100%	99.2%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DES	100%	100%	100%	98.2%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419

DMD	99.6%	99.3%	99.3%	74.1%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DNAJC19	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198
DOLK	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type Im, 610768
DOT1L	100%	100%	100%	98.3%	
DPM3	100%	100%	100%	98.3%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	100%	100%	100%	98.4%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956

DSC2	100%	100%	100%	99.4%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	100%	99.9%	100%	99.6%	Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193
DSP	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 8, 607450;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	98.5%	97.2%	100%	99.7%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169;Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis 2, 620971

DYRK1A	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 7, 614104
DZIP1	100%	100%	100%	99.7%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
EEF1A2	99.4%	96.8%	100%	97.6%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EHMT1	99.9%	99.7%	100%	98.9%	Kleefstra syndrome 1, 610253
EIF2AK4	100%	100%	100%	99.6%	Pulmonary venoocclusive disease 2, 234810
ELAC2	100%	100%	100%	99.4%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
ELN	100%	100%	100%	99.1%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
EMD	94.2%	89.4%	96.7%	63.6%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300

ENPP1	99.9%	99.3%	100%	99.6%	{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522
FAH	100%	100%	100%	99.4%	Tyrosinemia, type I, 276700
FBN1	100%	100%	100%	99.8%	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	99.2%	99.2%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBXO32	100%	100%	100%	99.1%	
FGF12	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 47, 617166

FHL1	100%	99.4%	98.4%	71.2%	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%	100%	99.4%	
FHOD3	100%	100%	100%	99.1%	Cardiomyopathy, familial hypertrophic, 28, 619402

FKRP	100%	100%	100%	96.9%	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%	100%	99.9%	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
FLII	100%	100%	100%	98.9%	Cardiomyopathy, dilated, 2J, 620635

FLNA	100%	99.8%	98.2%	67.3%	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%	100%	99.1%	Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524
FLT4	100%	100%	100%	99%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780

FNIP1	100%	100%	100%	99.7%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXC2	100%	100%	100%	94.5%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXH1	100%	100%	100%	99.1%	
FOXJ1	100%	100%	100%	98.2%	Ciliary dyskinesia, primary, 43, 618699
FOXL1	100%	100%	99.6%	92.6%	Otosclerosis 11, 620576
GAA	100%	100%	100%	99.3%	Glycogen storage disease II, 232300
GATA4	100%	100%	100%	97.7%	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%	99.9%	100%	98.4%	Congenital heart defects, multiple types, 5, 617912

GATA6	100%	100%	100%	95.2%	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%	100%	99.1%	?Cardiomyopathy, dilated, 2B, 614672
GATB	100%	100%	100%	99.5%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100%	100%	100%	98.9%	Combined oxidative phosphorylation deficiency 42, 618839
GBE1	100%	100%	100%	99.8%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GDF1	100%	99.7%	100%	97.8%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GDF2	100%	100%	100%	99.5%	Telangiectasia, hereditary hemorrhagic, type 5, 615506

GJA5	100%	100%	100%	99.6%	Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	91.4%	91.2%	99.2%	73.3%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLB1	100%	100%	100%	99.5%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLIS1	100%	100%	100%	98.9%	
GLYR1	100%	100%	100%	99.3%	
GMPPB	100%	100%	100%	99.4%	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%	100%	98.7%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
GNPTAB	100%	100%	100%	99.5%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GPD1L	100%	100%	100%	99.7%	Brugada syndrome 2, 611777
HADHA	100%	100%	100%	99.5%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	100%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HAND1	100%	100%	100%	97.5%	
HAND2	100%	100%	99.9%	89%	
HCN2	92.9%	88.3%	96.5%	86.4%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100%	100%	100%	98.8%	

HCN4	100%	100%	100%	98.3%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
HEY2	100%	100%	100%	96.9%	
HFE	100%	100%	100%	98.7%	Hemochromatosis, type 1, 235200
HJV	100%	100%	100%	99.5%	Hemochromatosis, type 2A, 602390
HSPB6	100%	100%	99.7%	95%	
HSPD1	99.6%	97.6%	100%	99.8%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
IDUA	100%	100%	100%	97.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
ILK	100%	100%	100%	99.1%	
ITGA7	100%	100%	100%	99.3%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	100%	100%	100%	99.5%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647

JAG1	100%	100%	100%	99.4%	?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%	100%	97.8%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873
JUP	100%	100%	100%	98.4%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
KANSL1	100%	100%	100%	99.7%	Koolen-De Vries syndrome, 610443
KBTBD13	100%	100%	100%	96.8%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA5	100%	100%	100%	98.8%	Atrial fibrillation, familial, 7, 612240
KCND2	100%	100%	100%	98.5%	
KCND3	100%	100%	100%	97.8%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNE1	100%	100%	100%	99.8%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695

KCNE2	100%	100%	100%	99.5%	Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493
KCNE3	100%	100%	100%	99.8%	?Brugada syndrome 6, 613119
KCNE4	100%	100%	100%	98.4%	
KCNE5	100%	99.5%	97.5%	64.3%	
KCNH2	100%	100%	100%	98.1%	Short QT syndrome 1, 609620;Long QT syndrome 2, 613688
KCNJ11	100%	100%	100%	99.2%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;{Diabetes mellitus, type 2, susceptibility to}, 125853;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ2	100%	100%	100%	99.2%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNJ5	100%	100%	100%	98.6%	Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677
KCNJ8	100%	100%	100%	99.7%	

KCNK3	100%	99.9%	100%	98.9%	Pulmonary hypertension, primary, 4, 615344
KCNN3	100%	100%	100%	98.5%	Zimmermann-Laband syndrome 3, 618658
KCNQ1	100%	99.7%	100%	98.1%	Short QT syndrome 2, 609621;Atrial fibrillation, familial, 3, 607554;Long QT syndrome 1, 192500;{Long QT syndrome 1, acquired, susceptibility to}, 192500;Jervell and Lange-Nielsen syndrome, 220400
KDR	100%	100%	100%	99.8%	{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089
KIF20A	100%	100%	100%	99.6%	?Cardiomyopathy, familial restrictive, 6, 619433
KLHL24	100%	100%	100%	99.9%	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294
KMT2A	99.2%	99.2%	100%	99.2%	Wiedemann-Steiner syndrome, 605130

KMT2D	100%	100%	100%	98.6%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KRAS	100%	100%	100%	99.8%	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%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855

LAMA4	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	85.3%	85.1%	98.7%	73.3%	Danon disease, 300257
LBX1	100%	100%	100%	98.4%	?Central hypoventilation syndrome, congenital, 3, 619483
LDB3	100%	100%	100%	98.7%	Left ventricular noncompaction 3, 601493;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LEFTY2	100%	100%	100%	98.8%	
LEMD2	100%	100%	100%	99%	Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500
LIMS2	100%	100%	100%	99.3%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMCD1	100%	100%	100%	99.2%	

LMNA	100%	100%	100%	98.7%	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%	100%	98.8%	Cardiomyopathy, dilated, 2G, 619897
LRRC10	100%	100%	100%	98.4%	
LZTR1	100%	100%	100%	99.1%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670

MED13L	100%	100%	100%	99.4%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MIB1	100%	100%	100%	99.6%	Left ventricular noncompaction 7, 615092
MIPEP	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 31, 617228
MLYCD	100%	100%	100%	98.9%	Malonyl-CoA decarboxylase deficiency, 248360
MMP21	100%	100%	100%	99.3%	Heterotaxy, visceral, 7, autosomal, 616749
MNS1	100%	100%	100%	99.7%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MTO1	94.7%	92.3%	100%	99.7%	Combined oxidative phosphorylation deficiency 10, 614702
MT-TI	99.6%	95.9%			
MUC16	100%	100%	100%	99.5%	
MYBPC3	100%	100%	100%	99%	Cardiomyopathy, hypertrophic, 4, 115197;Cardiomyopathy, dilated, 1MM, 615396;Left ventricular noncompaction 10, 615396
MYBPHL	100%	100%	100%	98.9%	

MYH11	100%	100%	100%	98.7%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYH6	100%	100%	100%	99.1%	?Atrial septal defect 3, 614089;{Sick sinus syndrome 3}, 614090;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251
MYH7	100%	100%	100%	99.2%	Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358
MYH7B	100%	100%	100%	98.8%	
MYL2	100%	100%	100%	99.2%	Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424

MYL3	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100%	100%	100%	98.7%	?Atrial fibrillation, familial, 18, 617280
MYL7	100%	100%	100%	99%	
MYLK3	100%	100%	100%	99.4%	
MYO6	100%	100%	100%	99.8%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821
MYOM1	100%	100%	100%	99.3%	
MYOT	100%	100%	100%	99.7%	Myopathy, myofibrillar, 3, 609200
MYOZ2	100%	100%	100%	99.6%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	98.4%	98.4%	100%	99.5%	Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248

MYRF	100%	100%	100%	98.5%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
MYZAP	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2K, 620894
NAA15	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NDUFAF1	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFB11	99%	93.8%	92%	56%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NEBL	100%	100%	100%	99.2%	
NEXN	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876

NF1	99.4%	99.4%	100%	99.7%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NKX2-5	100%	100%	100%	99%	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%	99.2%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NODAL	100%	100%	100%	98.9%	Heterotaxy, visceral, 5, 270100
NONO	96.2%	90.7%	99.1%	71.5%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOS1AP	100%	100%	100%	98.8%	Nephrotic syndrome, type 22, 619155

NOTCH1	99.4%	99.1%	100%	98.8%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	100%	100%	100%	99.7%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NPPA	100%	100%	100%	99%	Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201
NPPB	100%	100%	100%	99.6%	
NR2F2	100%	100%	100%	98.1%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NRAP	100%	100%	100%	99.4%	
NSD1	100%	100%	100%	99.5%	Sotos syndrome, 117550
NUP155	100%	100%	100%	99.7%	?Atrial fibrillation 15, 615770
ODAD1	100%	100%	100%	98.6%	Ciliary dyskinesia, primary, 20, 615067
PCCA	100%	100%	100%	99.8%	Propionicacidemia, 606054
PCCB	99.2%	96.1%	100%	99.7%	Propionicacidemia, 606054
PDLIM3	100%	100%	100%	99.8%	
PDLIM5	99.5%	97.3%	100%	99.3%	

PEX5	100%	100%	100%	98.5%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	97.9%	97.9%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PGM1	94%	94%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PHKA1	100%	99.8%	99.3%	74.3%	Muscle glycogenosis, 300559
PHYH	100%	100%	100%	99.2%	Refsum disease, 266500
PITX2	100%	100%	100%	97.9%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PKD1L1	100%	100%	100%	99.4%	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	99%	97.1%	100%	99.3%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLD1	100%	100%	100%	99.7%	Cardiac valvular dysplasia 1, 212093

PLEKHM2	100%	100%	100%	98.8%	
PLN	100%	100%	100%	99.1%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874
PLXND1	100%	99.9%	100%	98.7%	Congenital heart defects, multiple types, 9, 620294
PMM2	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	100%	100%	100%	98.6%	Neutral lipid storage disease with myopathy, 610717
POMT1	100%	100%	100%	99%	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%	99.9%	98.1%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156
PPA2	100%	100%	100%	99.5%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PPCDC	100%	100%	100%	99.4%	
PPCS	100%	100%	100%	99.2%	Cardiomyopathy, dilated, 2C, 618189
PPP1R13L	100%	100%	100%	96.2%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519
PRDM16	100%	100%	100%	97.8%	Left ventricular noncompaction 8, 615373;Cardiomyopathy, dilated, 1LL, 615373
PRDM6	100%	100%	99.9%	96.3%	Patent ductus arteriosus 3, 617039

PRKAG2	100%	100%	100%	98.7%	Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858
PRKD1	100%	100%	99.9%	97.2%	Congenital heart defects and ectodermal dysplasia, 617364
PTPN11	89.7%	89.2%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
QRSL1	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 40, 618835
RAF1	96.6%	93.5%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RANGRF	100%	100%	100%	99.3%	
RBCK1	100%	100%	100%	97.5%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX2	92.6%	91.1%	100%	99.1%	
RBM20	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 1DD, 613172

RIT1	100%	100%	100%	99%	Noonan syndrome 8, 615355
ROBO4	100%	100%	100%	98.9%	Aortic valve disease 3, 618496
RPL3L	100%	100%	100%	97.3%	Cardiomyopathy, dilated, 2D, 619371
RPS6KB1	100%	100%	100%	99.2%	
RRAD	100%	100%	100%	95.4%	
RRAGC	100%	100%	100%	99.3%	Long-Olsen-Distelmaier syndrome, 620609
RYR2	100%	100%	100%	99.6%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000
SCN10A	100%	100%	100%	99.5%	Episodic pain syndrome, familial, 2, 615551
SCN1B	100%	99.1%	100%	99.1%	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%	98.8%	Atrial fibrillation, familial, 14, 615378
SCN3B	100%	100%	100%	99.1%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4B	100%	100%	100%	98.3%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN5A	100%	100%	100%	99.2%	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;{Sudden infant death syndrome, susceptibility to}, 272120
SCO2	100%	100%	100%	99.4%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377

SDHA	100%	100%	100%	99.4%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma /paraganglioma syndrome 5, 614165
SGCA	100%	100%	100%	98.8%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100%	100%	100%	99%	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHMT2	100%	100%	100%	99.3%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC2	100%	100%	100%	99.7%	Noonan syndrome-like with loose anagen hair 1, 607721

SHOX2	100%	100%	99.9%	96.8%	
SHROOM3	100%	100%	100%	99.2%	
SLC22A5	100%	100%	100%	99.2%	Carnitine deficiency, systemic primary, 212140
SLC25A20	100%	100%	100%	99.3%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	100%	100%	100%	98.8%	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%	100%	98.8%	
SLC4A3	100%	100%	100%	98.8%	Short QT syndrome 7, 620231
SLC6A6	100%	100%	100%	99%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLMAP	100%	100%	100%	99.6%	
SMAD1	100%	100%	100%	99.7%	
SMAD5	100%	100%	100%	99.9%	

SMAD6	100%	100%	100%	96.5%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
SMARCA4	100%	100%	100%	98.7%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SNTA1	100%	100%	100%	98.8%	Long QT syndrome 12, 612955
SOD2	100%	100%	100%	99.3%	{Microvascular complications of diabetes 6}, 612634
SOS1	98.7%	98.3%	100%	99.7%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOX7	100%	100%	100%	96%	
SRF	100%	100%	100%	96.6%	
SRI	100%	100%	100%	99.6%	
SURF1	100%	100%	100%	98.5%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SVIL	100%	100%	100%	99.5%	Myofibrillar myopathy 10, 619040
TAB2	100%	100%	100%	98.8%	Congenital heart defects, nonsyndromic, 2, 614980

TAF1	98.7%	98.4%	98.7%	70.9%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TAF1A	100%	100%	100%	99.8%	
TAFAZZIN	100%	99.6%	97.2%	66.7%	Barth syndrome, 302060
TBX1	96.8%	93%	99.7%	89.1%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX20	100%	100%	100%	99.1%	Atrial septal defect 4, 611363
TBX5	100%	100%	100%	99.1%	Holt-Oram syndrome, 142900
TCAP	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TDGF1	100%	100%	100%	99.8%	
TECRL	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	100%	100%	100%	97.8%	Patent ductus arteriosus 2, 617035;Char syndrome, 169100

TGFB3	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
THBS4	100%	100%	100%	99.4%	
TJP1	100%	100%	100%	99.6%	
TLL1	100%	100%	100%	99.6%	Atrial septal defect 6, 613087
TMEM260	100%	100%	100%	99.6%	Structural heart defects and renal anomalies syndrome, 617478
TMEM43	100%	100%	100%	99%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMPO	100%	100%	100%	99.4%	
TNNC1	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	100%	100%	100%	97.5%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286

TNNI3K	100%	100%	100%	99.6%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	100%	100%	100%	99.1%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494
TNS1	100%	100%	100%	99.2%	
TOR1AIP1	100%	100%	100%	99.1%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TPM1	100%	100%	100%	98.9%	Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878
TRDN	100%	100%	100%	99.7%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TRIM63	100%	100%	100%	99.4%	
TRPM4	100%	100%	100%	98.5%	Progressive familial heart block, type IB, 604559;Erythrokeratodermia variabilis et progressiva 6, 618531

TSC1	100%	100%	100%	99.3%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSFM	94.3%	94.3%	100%	98.9%	Combined oxidative phosphorylation deficiency 3, 610505
TTN	100%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Cardiomyopathy, familial hypertrophic, 9, 613765;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;Myopathy, myofibrillar, 9, with early respiratory failure, 603689
TTR	100%	100%	100%	99.6%	Amyloidosis, hereditary, transthyretin-related, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
TULP3	100%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902

TXNRD2	100%	100%	100%	98.8%	?Glucocorticoid deficiency 5, 617825
VCL	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255
VEZF1	100%	100%	100%	98.3%	?Cardiomyopathy, dilated, 1OO, 620247
XIRP2	100%	100%	100%	99.7%	
XK	100%	98.9%	99.2%	70.9%	McLeod syndrome, 300842
ZBTB17	100%	100%	100%	99.4%	
ZFPM2	100%	100%	100%	99.5%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZIC3	100%	100%	98.2%	68.6%	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.
TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.
TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.
srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 17th, 2023.
This list is accurate for panel version DG 4.0.0

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors