HEART DISORDERS PANEL[1] DG-5.1.0 (392 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
AARS2	100%	100%	100%	100%	99.5%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABCC6	94.4%	94.4%	100%	100%	99.5%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	96%	96%	100%	100%	99.8%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABL1	100%	100%	100%	100%	99.4%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACAD8	100%	99.9%	100%	99.9%	99.5%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	100%	100%	100%	100%	99.3%	VLCAD deficiency, 201475

ACSF3	94.7%	94.7%	100%	100%	99.3%	Combined malonic and methylmalonic aciduria, 614265
ACTA1	100%	100%	100%	99.8%	97.9%	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%	100%	99.8%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042
ACTC1	90.7%	90.6%	100%	100%	99.6%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACTN2	99.8%	98.4%	100%	99.8%	98.6%	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%	100%	99.4%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS10	100%	100%	100%	100%	99.6%	Weill-Marchesani syndrome 1, recessive, 277600

ADAMTS17	100%	100%	100%	100%	99.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS19	94%	94%	100%	99.9%	99.3%	Cardiac valvular dysplasia 2, 620067
ADAMTSL2	100%	100%	100%	99.9%	99.4%	Geleophysic dysplasia 1, 231050
ADCY5	97.5%	96.9%	100%	99.9%	99.1%	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%	100%	99.7%	Helsmoortel-van der Aa syndrome, 615873
AEBP1	100%	100%	100%	99.9%	99.3%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AGK	91.6%	88.9%	100%	100%	100%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGL	100%	100%	100%	100%	99.7%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400
AGPAT2	100%	99.9%	100%	100%	98.9%	Lipodystrophy, congenital generalized, type 1, 608594
AKAP9	99.6%	99.6%	100%	100%	99.7%	?Long QT syndrome 11, 611820
ALDH1A2	88.9%	88.9%	100%	100%	99.5%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ALMS1	100%	100%	100%	100%	99.8%	Alstrom syndrome, 203800

ALPK3	100%	100%	100%	100%	99.5%	Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	100%	100%	100%	100%	99.7%	Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919
ANKRD1	100%	100%	100%	99.9%	99%	
ANKRD11	100%	100%	100%	100%	99.2%	KBG syndrome, 148050
ATPAF2	100%	100%	100%	100%	99.5%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	100%	100%	100%	100%	99.5%	?Neuronopathy, distal hereditary motor, autosomal dominant 15, 621094;Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954;Charcot-Marie-Tooth disease, axonal, type 2JJ, 621095
BAG5	100%	100%	100%	100%	98.8%	Cardiomyopathy, dilated, 2F, 619747
BANF1	100%	100%	100%	99.8%	98.5%	Nestor-Guillermo progeria syndrome, 614008
BCL9L	100%	100%	100%	100%	99.3%	
BICD2	100%	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
BMP10	100%	100%	100%	100%	99.9%	

BMPR2	100%	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%	100%	99.8%	98.2%	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%	100%	99.6%	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
CACNA1C	100%	100%	100%	100%	99.5%	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%	100%	99.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 110, 620149
CACNB2	100%	100%	100%	100%	99.3%	Brugada syndrome 4, 611876
CALM1	100%	100%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100%	99.9%	98.9%	Long QT syndrome 15, 616249
CALM3	100%	100%	100%	100%	99.7%	Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CAP2	82%	82%	100%	99.9%	99.8%	Cardiomyopathy, dilated, 2I, 620462
CASQ2	87.5%	87.5%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASZ1	100%	99.6%	100%	99.9%	98.7%	
CAV1	64.8%	64.8%	100%	100%	98.8%	Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721

CAV3	100%	100%	100%	100%	99.2%	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%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDK13	100%	100%	100%	100%	99.1%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CFAP45	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP52	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376
CHD4	100%	100%	100%	100%	99.5%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800

CHKB	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	100%	100%	100%	100%	99.9%	
CIROP	98.3%	93.9%	100%	100%	99.7%	Heterotaxy, visceral, 12, autosomal, 619702
CIROZ	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 14, autosomal, 621080
CITED2	100%	100%	100%	99.8%	98.5%	Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431
COL3A1	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ7	100%	100%	100%	100%	99.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
CORIN	97.6%	97.6%	100%	100%	99.8%	?Cardiomyopathy, familial hypertrophic, 30, atrial, 620734;Preeclampsia/eclampsia 5, 614595
COX15	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CPT1A	100%	100%	100%	100%	99.5%	CPT deficiency, hepatic, type IA, 255120

CPT2	97.3%	96.3%	100%	100%	99.7%	{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%	100%	99.6%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPTO	100%	100%	100%	100%	99.9%	
CRYAB	100%	100%	100%	100%	99.7%	Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
CSRP3	100%	100%	100%	100%	99.9%	?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124
CTNNA3	100%	100%	100%	100%	100%	Arrhythmogenic right ventricular dysplasia 13, 615616
CTNND1	100%	100%	100%	100%	99.9%	Blepharocheilodontic syndrome 2, 617681
DCHS1	100%	100%	100%	100%	99.6%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390

DES	92.6%	92.6%	100%	99.9%	98.6%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DMD	99.6%	99.6%	99.2%	91.5%	73.3%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DNAH5	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 40, 618300
DNAJC19	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198
DOCK6	100%	100%	100%	99.9%	99.3%	Adams-Oliver syndrome 2, 614219
DOLK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Im, 610768
DOT1L	100%	100%	100%	100%	99.3%	Nil-Deshwar neurodevelopmental syndrome, 621265
DPM3	100%	100%	100%	100%	98.8%	?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%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	100%	100%	100%	99.9%	99.5%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	88%	88%	100%	100%	99.7%	Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193
DSP	99.4%	99.2%	100%	100%	99.5%	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	98.7%	97.2%	100%	100%	99.8%	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
DVL3	100%	100%	100%	99.9%	99.1%	Robinow syndrome, autosomal dominant 3, 616894

DYRK1A	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 7, 614104
DZIP1	100%	100%	100%	100%	99.7%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
EEF1A2	99.4%	95.6%	100%	100%	99.1%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EHMT1	97.7%	97.7%	100%	100%	99.7%	Kleefstra syndrome 1, 610253
EIF2AK4	99%	99%	100%	100%	99.8%	Pulmonary venoocclusive disease 2, 234810
EIF3A	100%	100%	100%	99.9%	99.5%	
EIF3B	100%	100%	100%	100%	99.2%	
ELAC2	94.8%	94.8%	100%	99.9%	99.5%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
ELN	100%	99.7%	100%	99.9%	99.1%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
EMD	93.1%	91.5%	97.5%	87.2%	69.2%	Cardiomyopathy, dilated, 3C, 301163;Emery-Dreifuss muscular dystrophy 1, X-linked, 310300

ENPP1	100%	100%	100%	100%	99.8%	{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
ETS1	100%	100%	100%	100%	99.7%	
FAH	95.2%	93.3%	100%	100%	99.5%	Tyrosinemia, type I, 276700
FBN1	100%	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%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBXO32	100%	100%	100%	100%	99.6%	
FGF12	100%	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 47, 617166
FGF8	100%	100%	100%	99.8%	98.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

FHL1	100%	100%	99.4%	91.2%	73.3%	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	91.7%	91.7%	100%	100%	99.7%	
FHOD3	97.6%	97.6%	100%	100%	99.5%	Cardiomyopathy, familial hypertrophic, 28, 619402
FKRP	100%	100%	100%	100%	99.3%	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	95.2%	94.9%	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%	100%	99.5%	Cardiomyopathy, dilated, 2J, 620635
FLNA	100%	100%	98.3%	87.4%	68.7%	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%	100%	99.6%	Cardiomyopathy, familial dilated, 1PP, 617047;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	99.1%	97.6%	100%	100%	99.4%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FNIP1	100%	100%	100%	99.9%	99.7%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXC2	100%	100%	100%	99.8%	98.2%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXH1	100%	100%	100%	100%	99.5%	
FOXJ1	100%	100%	100%	99.9%	99.4%	Ciliary dyskinesia, primary, 43, 618699
GAA	100%	100%	100%	100%	99.5%	Pompe disease, late-onset, 621314;Pompe disease, infantile-onset, 232300

GATA4	100%	100%	100%	99.9%	98.2%	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%	99.1%	100%	99.9%	98.6%	Congenital heart defects, multiple types, 5, 617912
GATA6	93.8%	93.8%	100%	99.8%	98.1%	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%	99.9%	99.2%	?Cardiomyopathy, dilated, 2B, 614672
GATB	100%	100%	100%	100%	99.7%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	80.1%	80.1%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 42, 618839
GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GDF1	100%	100%	100%	100%	98.9%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GDF2	100%	100%	100%	100%	99.1%	Telangiectasia, hereditary hemorrhagic, type 5, 615506

GET3	87.9%	87.9%	100%	99.9%	98.9%	?Cardiomyopathy, dilated, 2H, 620203
GJA5	100%	100%	100%	100%	99.9%	Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	91.4%	91.4%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLB1	100%	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%	100%	99%	
GLYR1	100%	100%	100%	100%	99.2%	
GMPPB	100%	100%	100%	100%	99.7%	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%	100%	99.6%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
GNPTAB	100%	100%	100%	100%	99.8%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500

GPD1L	100%	100%	100%	100%	99.4%	Brugada syndrome 2, 611777
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HAND1	100%	100%	100%	100%	98.9%	
HAND2	100%	100%	100%	99.4%	94%	
HCN2	95.1%	93.5%	99.3%	97.5%	93%	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%	99.9%	99.1%	
HCN4	100%	100%	100%	100%	99%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
HECTD1	98.5%	98.5%	100%	100%	99.8%	
HEY2	100%	100%	100%	99.9%	98.8%	
HFE	100%	99.7%	100%	100%	99.4%	Hemochromatosis, type 1, 235200
HJV	100%	100%	100%	100%	99.8%	Hemochromatosis, type 2A, 602390
HSPB6	100%	100%	100%	99.6%	97.6%	

HSPD1	99.9%	98.9%	100%	100%	99.9%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
IDUA	96%	96%	100%	99.8%	98.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
ILK	100%	100%	100%	100%	99.5%	
ISL1	100%	100%	100%	100%	99.8%	
ITGA7	100%	99.9%	100%	100%	99.5%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	76.9%	76.9%	100%	99.9%	99.4%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
JAG1	100%	100%	100%	100%	99.6%	?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	92.6%	92.6%	100%	99.9%	98.5%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873
JUP	100%	100%	100%	100%	99.5%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
KANSL1	98.1%	98.1%	100%	100%	99.8%	Koolen-De Vries syndrome, 610443

KAT6B	100%	100%	100%	100%	99.5%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KBTBD13	100%	100%	100%	100%	99.1%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 32, 616366
KCNA5	100%	100%	100%	99.9%	99.3%	Atrial fibrillation, familial, 7, 612240
KCND2	100%	100%	100%	99.9%	99.7%	
KCND3	100%	100%	100%	99.9%	99%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNE1	100%	100%	100%	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNE2	100%	100%	100%	100%	99.7%	Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493
KCNE3	100%	100%	100%	100%	99.8%	?Brugada syndrome 6, 613119
KCNE4	100%	100%	100%	100%	99.4%	
KCNE5	100%	100%	99.1%	90.7%	71.9%	
KCNH2	100%	100%	100%	100%	99%	Short QT syndrome 1, 609620;Long QT syndrome 2, 613688

KCNJ11	100%	100%	100%	100%	99.5%	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%	99.8%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNJ5	100%	100%	100%	100%	99.2%	Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677
KCNJ8	89.3%	89.3%	100%	100%	99.7%	
KCNK3	100%	100%	100%	100%	99%	Developmental delay with sleep apnea, 621402;Pulmonary hypertension, primary, 4, 615344
KCNN3	100%	100%	100%	99.9%	99%	Zimmermann-Laband syndrome 3, 618658
KCNQ1	100%	100%	100%	99.9%	99.3%	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%	100%	99.7%	{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089

KIF20A	100%	100%	100%	100%	99.7%	?Cardiomyopathy, familial restrictive, 6, 619433
KLF13	100%	100%	99.9%	99.2%	96.1%	
KLHL24	91.5%	91.3%	100%	100%	99.7%	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%	100%	99.5%	Wiedemann-Steiner syndrome, 605130
KMT2D	100%	100%	100%	100%	99.2%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920

KRAS	100%	100%	100%	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%	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%	100%	99.9%	Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	70.7%	70.7%	98.9%	91%	71.6%	Danon disease, 300257
LBX1	100%	100%	100%	100%	99.1%	?Central hypoventilation syndrome, congenital, 3, 619483

LDB3	95.9%	94.9%	100%	100%	99.4%	Left ventricular noncompaction 3, 601493;Cardiomyopathy, dilated, 2L, 621237;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LEMD2	100%	100%	100%	100%	99.8%	Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500
LIMS2	100%	99.9%	100%	100%	99.8%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMCD1	100%	100%	100%	99.9%	99.3%	
LMNA	100%	100%	100%	99.9%	99.3%	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%	100%	99.2%	Cardiomyopathy, dilated, 2G, 619897

LOX	100%	100%	100%	99.9%	98.5%	Aortic aneurysm, familial thoracic 10, 617168
LRRC10	100%	100%	100%	100%	99.8%	
LTBP2	100%	100%	100%	100%	99.7%	Glaucoma 3, primary congenital, D, 613086;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;?Weill-Marchesani syndrome 3, recessive, 614819
LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MED13L	100%	100%	100%	100%	99.6%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MEGF8	100%	100%	100%	99.8%	98.9%	Carpenter syndrome 2, 614976
MEIS2	89.6%	88.5%	100%	100%	99.6%	Cleft palate, cardiac defects, and impaired intellectual development, 600987
MESP1	100%	100%	100%	99.9%	99.3%	
MGRN1	100%	100%	100%	100%	99.5%	
MIB1	94.8%	94.8%	100%	100%	99.7%	Left ventricular noncompaction 7, 615092
MIPEP	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 31, 617228
MLYCD	100%	100%	100%	100%	99.4%	Malonyl-CoA decarboxylase deficiency, 248360

MMP21	100%	100%	100%	100%	99.4%	Heterotaxy, visceral, 7, autosomal, 616749
MNS1	100%	100%	100%	99.9%	99.8%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MRPL44	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 16, 615395
MST1R	100%	100%	100%	100%	99.6%	{Nasopharyngeal carcinoma, susceptibility to, 3}, 617075
MTO1	94.8%	91.9%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 10, 614702
MT-TI	99.6%	95.9%				
MUC16	100%	100%	100%	100%	99.6%	
MYBPC3	100%	100%	100%	100%	99.2%	Cardiomyopathy, hypertrophic, 4, 115197;Cardiomyopathy, dilated, 1MM, 615396;Left ventricular noncompaction 10, 615396
MYBPHL	99.9%	98.7%	100%	100%	99.4%	
MYH11	100%	100%	100%	100%	99.3%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYH6	100%	100%	100%	100%	99.4%	?Atrial septal defect 3, 614089;{Sick sinus syndrome 3}, 614090;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251

MYH7	99.1%	99.1%	100%	100%	99.7%	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%	100%	99.2%	
MYL2	100%	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	81.6%	79.9%	100%	100%	99.2%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100%	100%	100%	99.9%	99.5%	?Atrial fibrillation, familial, 18, 617280
MYL7	100%	100%	100%	100%	99.7%	
MYLK3	100%	100%	100%	100%	99.8%	
MYO5B	100%	100%	100%	100%	99.5%	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868
MYO6	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821

MYOM1	100%	100%	100%	100%	99.7%	
MYOT	100%	100%	100%	100%	99.8%	Myopathy, myofibrillar, 3, 609200
MYOZ2	88.7%	88.7%	100%	100%	99.9%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	98.4%	98.4%	100%	100%	99.6%	Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248
MYRF	100%	99.8%	100%	99.9%	99.2%	Nanophthalmos 1, 600165;Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
MYZAP	80%	80%	100%	99.9%	99.7%	Cardiomyopathy, dilated, 2K, 620894
NAA15	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NDUFAF1	91.6%	91.5%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFB11	99.9%	99.3%	93.6%	79.3%	59.7%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NEBL	100%	100%	100%	100%	99.5%	

NEXN	100%	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 2M, autosomal recessive, 621261;Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876
NF1	99.4%	99.4%	100%	100%	99.8%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NKX2-5	100%	100%	100%	99.7%	98.1%	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%	99.5%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NODAL	100%	100%	100%	99.9%	99.8%	Heterotaxy, visceral, 5, autosomal, 270100
NONO	94.7%	89.1%	99.4%	92.9%	75.5%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOS1AP	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 22, 619155

NOTCH1	99.1%	99%	100%	99.9%	99.4%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	100%	100%	100%	100%	99.8%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NPHP4	100%	100%	100%	100%	99.5%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NPPA	100%	100%	100%	100%	99.7%	Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201
NPPB	100%	100%	100%	100%	100%	
NR2F2	100%	100%	100%	100%	99.4%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NRAP	100%	100%	100%	100%	99.7%	
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
NUP155	93.6%	93.6%	100%	100%	99.8%	?Atrial fibrillation 15, 615770
ODAD1	100%	100%	100%	99.9%	99.4%	Ciliary dyskinesia, primary, 20, 615067
ODAD2	96.1%	96.1%	100%	100%	99.7%	Ciliary dyskinesia, primary, 23, 615451
OLA1	100%	100%	100%	100%	99.8%	
PBX1	100%	100%	100%	100%	99.7%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	84.6%	84.4%	100%	100%	99.7%	Propionicacidemia, 606054
PCCB	86.8%	85.3%	100%	100%	99.8%	Propionicacidemia, 606054

PDLIM3	100%	100%	100%	100%	99.6%	
PDLIM5	99.9%	98.2%	100%	100%	99.8%	
PEX5	100%	100%	100%	99.8%	99%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PHKA1	100%	100%	99.5%	91.9%	72.6%	Muscle glycogenosis, 300559
PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500
PITX2	100%	100%	100%	100%	99.1%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PKD1L1	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	98.9%	98.1%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLD1	100%	100%	100%	100%	99.8%	Cardiac valvular dysplasia 1, 212093
PLEKHM2	96.7%	96.7%	100%	100%	99.5%	

PLN	100%	100%	100%	100%	99.9%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874
PLXND1	100%	100%	100%	99.9%	99.2%	Congenital heart defects, multiple types, 9, 620294
PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	80.4%	80.4%	100%	100%	99.6%	Neutral lipid storage disease with myopathy, 610717
POMT1	100%	99.7%	100%	100%	99.3%	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	94.9%	94.9%	100%	99.8%	98.4%	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
POPDC1	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812

POPDC2	100%	100%	100%	100%	99.7%	Cardiac conduction disease with or without cardiomyopathy 2, 621367
PPA2	82.6%	82.6%	100%	100%	99.7%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PPCDC	100%	100%	100%	100%	100%	
PPCS	100%	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2C, 618189
PPP1R13L	100%	100%	100%	99.9%	98.5%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519
PRDM16	100%	100%	100%	99.9%	98.9%	Left ventricular noncompaction 8, 615373;Cardiomyopathy, dilated, 1LL, 615373
PRDM6	100%	100%	100%	99.9%	98.8%	Patent ductus arteriosus 3, 617039
PRKAG2	100%	100%	100%	99.9%	99.6%	Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858
PRKD1	100%	100%	100%	99.8%	98.9%	Congenital heart defects and ectodermal dysplasia, 617364
PTPN11	89.2%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PUF60	100%	100%	100%	100%	99.2%	Verheij syndrome, 615583

QRSL1	91.9%	89.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 40, 618835
RAF1	96.1%	93.8%	100%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RANGRF	100%	100%	100%	99.9%	99.2%	
RBCK1	100%	100%	100%	99.8%	98.8%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX2	85.5%	84.7%	100%	100%	99.6%	
RBM20	100%	100%	100%	100%	99.4%	Cardiomyopathy, dilated, 1DD, 613172
RIT1	100%	100%	100%	100%	99.8%	Noonan syndrome 8, 615355
ROBO4	100%	100%	100%	100%	99.4%	Aortic valve disease 3, 618496
RPL3L	100%	99.9%	100%	99.8%	99.2%	Cardiomyopathy, dilated, 2D, 619371
RPS6KB1	100%	100%	100%	100%	99.5%	
RRAD	100%	100%	100%	100%	99.4%	
RRAGC	100%	100%	100%	100%	99.5%	Long-Olsen-Distelmaier syndrome, 620609
RYR2	99.8%	99.8%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000
SCN10A	100%	100%	100%	100%	99.8%	Episodic pain syndrome, familial, 2, 615551

SCN1B	100%	100%	100%	100%	99.5%	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%	99.9%	99%	Atrial fibrillation, familial, 14, 615378
SCN3B	90%	89.7%	100%	100%	99.7%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4B	100%	100%	100%	100%	99.2%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN5A	100%	100%	100%	100%	99.5%	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%	100%	99.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377

SDHA	92.6%	89.3%	100%	100%	99.7%	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
SECISBP2	100%	100%	100%	100%	99.7%	Thyroid hormone metabolism, abnormal, 1, 609698
SF3B2	100%	100%	100%	100%	99.4%	Craniofacial microsomia, 164210
SGCA	97.5%	97.5%	100%	100%	99.2%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94%	94%	100%	100%	99.7%	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	83.2%	83.2%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHMT2	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC2	100%	100%	100%	100%	99.9%	Noonan syndrome-like with loose anagen hair 1, 607721
SHOX2	100%	100%	99.9%	99.3%	97.2%	
SHROOM3	100%	100%	100%	100%	99.6%	

SLC22A5	97.1%	97.1%	100%	100%	99.5%	Carnitine deficiency, systemic primary, 212140
SLC25A20	100%	100%	100%	100%	99.4%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A4	100%	100%	100%	100%	99.4%	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%	100%	99.5%	
SLC4A3	100%	100%	100%	100%	99.6%	Short QT syndrome 7, 620231
SLC6A6	100%	100%	100%	100%	99.4%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLMAP	100%	100%	100%	100%	99.8%	
SMAD1	100%	100%	100%	100%	99.7%	
SMAD2	86.7%	86.7%	100%	100%	99.8%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD5	100%	100%	100%	100%	99.9%	
SMAD6	100%	100%	100%	99.8%	98.1%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439

SMARCA4	100%	100%	100%	99.9%	99.3%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SNTA1	92.9%	92.9%	100%	100%	99.3%	Long QT syndrome 12, 612955
SOD2	100%	100%	100%	100%	99.8%	{Microvascular complications of diabetes 6}, 612634
SOS1	98.8%	98.6%	100%	100%	99.6%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOX7	100%	100%	100%	100%	98.8%	
SRF	99.7%	97.4%	100%	99.9%	98.9%	
SRI	100%	100%	100%	100%	99.6%	
STRA6	100%	100%	100%	100%	99.3%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
SURF1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SVIL	100%	100%	100%	100%	99.4%	Myofibrillar myopathy 10, 619040
TAB2	100%	100%	100%	100%	98.7%	Congenital heart defects, nonsyndromic, 2, 614980
TAF1	98.7%	98.7%	98.9%	89%	70.8%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TAF1A	91.1%	90.8%	100%	100%	99.9%	
TAFAZZIN	100%	100%	98.6%	89.7%	70.1%	Barth syndrome, 302060

TAX1BP3	100%	100%	100%	100%	99.2%	
TBX1	98.6%	95.7%	100%	99.4%	96.1%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX20	100%	100%	100%	99.9%	99.4%	Atrial septal defect 4, 611363
TBX5	100%	100%	100%	100%	99.4%	Holt-Oram syndrome, 142900
TCAP	100%	100%	100%	100%	99.7%	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TECRL	78%	78%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TFAP2B	100%	100%	100%	100%	99.4%	Patent ductus arteriosus 2, 617035;Char syndrome, 169100
TGFB3	88.3%	88.3%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
THBS4	100%	100%	100%	100%	99.7%	
TJP1	100%	100%	100%	100%	99.7%	
TLL1	100%	100%	100%	100%	99.8%	Atrial septal defect 6, 613087
TMEM260	94.8%	94.8%	100%	100%	99.7%	Structural heart defects and renal anomalies syndrome, 617478

TMEM43	100%	100%	100%	100%	99.5%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMPO	100%	100%	100%	100%	99.8%	
TNNC1	100%	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	100%	100%	100%	100%	98.2%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	100%	100%	100%	100%	99.8%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	100%	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494
TNS1	100%	100%	100%	100%	99.4%	
TOP3A	95.7%	95%	100%	100%	99.5%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098

TOR1AIP1	100%	100%	100%	100%	99.8%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TPM1	91.3%	91.3%	100%	100%	99.5%	Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878
TRDN	92.7%	92.7%	100%	100%	99.8%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TRIM63	100%	100%	100%	100%	99.7%	Cardiomyopathy, familial hypertrophic, 31, 621270
TRPM4	100%	100%	100%	100%	99.4%	Progressive familial heart block, type IB, 604559;Erythrokeratodermia variabilis et progressiva 6, 618531
TSC1	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSFM	91.5%	91.1%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 3, 610505

TTN	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;?Cardiomyopathy, familial hypertrophic, 9, 613765;Myopathy myofibrillar, 9, with early respiratory failure, 603689
TTR	100%	100%	100%	100%	99.5%	Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
TULP3	93.4%	93.4%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902
TXNRD2	100%	100%	100%	100%	99.4%	?Glucocorticoid deficiency 5, 617825
UNC45B	96.1%	95.9%	100%	100%	99.7%	?Cataract 43, 616279;Myofibrillar myopathy 11, 619178
VCL	93.7%	93.6%	100%	100%	99.6%	Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255
VEZF1	86%	86%	100%	99.9%	99.4%	?Cardiomyopathy, dilated, 1OO, 620247
XIRP2	100%	100%	100%	100%	99.8%	
XK	100%	99.9%	99.4%	92.2%	74%	McLeod syndrome, 300842
ZBTB17	100%	100%	100%	100%	99.6%	

ZFPM2	100%	100%	100%	100%	99.8%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZIC3	100%	100%	98.4%	87.4%	68.6%	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390

Gene symbols used follow HGNC 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 mapped against GRCh38.
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 mapped against GRCh38.
srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS 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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors