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HEART DISORDERS PANEL[1] DG-5.2.0 (393 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	n.t.b					Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABCC6	n.t.b					Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	n.t.b					Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABL1	n.t.b					Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACAD8	n.t.b					Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	n.t.b					Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADVL	n.t.b					VLCAD deficiency, 201475

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ACSF3	n.t.b					Combined malonic and methylmalonic aciduria, 614265
ACTA1	n.t.b					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	n.t.b					Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042
ACTC1	n.t.b					Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACTN2	n.t.b					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	n.t.b					Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS10	n.t.b					Weill-Marchesani syndrome 1, recessive, 277600

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ADAMTS17	n.t.b					Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS19	n.t.b					Cardiac valvular dysplasia 2, 620067
ADAMTSL2	n.t.b					Geleophysic dysplasia 1, 231050
ADCY5	n.t.b					Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADNP	n.t.b					Helsmoortel-van der Aa syndrome, 615873
AEBP1	n.t.b					Ehlers-Danlos syndrome, classic-like, 2, 618000
AGK	n.t.b					Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGL	n.t.b					Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400
AGPAT2	n.t.b					Lipodystrophy, congenital generalized, type 1, 608594
AHNAK	n.t.b					
AKAP9	n.t.b					?Long QT syndrome 11, 611820
ALDH1A2	n.t.b					Diaphragmatic hernia 4, with cardiovascular defects, 620025

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ALMS1	n.t.b					Alstrom syndrome, 203800
ALPK3	n.t.b					Cardiomyopathy, familial hypertrophic 27, 618052
ANK2	n.t.b					Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919
ANKRD1	n.t.b					
ANKRD11	n.t.b					KBG syndrome, 148050
ATPAF2	n.t.b					?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BAG3	n.t.b					?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	n.t.b					Cardiomyopathy, dilated, 2F, 619747
BANF1	n.t.b					Nestor-Guillermo progeria syndrome, 614008
BCL9L	n.t.b					
BICD2	n.t.b					Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290

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BMP10	n.t.b					
BMPR2	n.t.b					Pulmonary hypertension, familial primary, 1, with or without HHT, 178600;Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600;Pulmonary venoocclusive disease 1, 265450
BRAF	n.t.b					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	n.t.b					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

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CACNA1C	n.t.b					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	n.t.b					Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA2D1	n.t.b					Developmental and epileptic encephalopathy 110, 620149
CACNB2	n.t.b					Brugada syndrome 4, 611876
CALM1	n.t.b					Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247
CALM2	n.t.b					Long QT syndrome 15, 616249
CALM3	n.t.b					Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CAP2	n.t.b					Cardiomyopathy, dilated, 2I, 620462
CASQ2	n.t.b					Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASZ1	n.t.b					

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CAV1	n.t.b					Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721
CAV3	n.t.b					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	n.t.b					Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDK13	n.t.b					Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CFAP45	n.t.b					Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP52	n.t.b					Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	n.t.b					Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	n.t.b					Heterotaxy, visceral, 2, autosomal, 605376

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CHD4	n.t.b					Sifrim-Hitz-Weiss syndrome, 617159
CHD7	n.t.b					Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHKB	n.t.b					Muscular dystrophy, congenital, megaconial type, 602541
CHRM2	n.t.b					
CIROP	n.t.b					Heterotaxy, visceral, 12, autosomal, 619702
CIROZ	n.t.b					Heterotaxy, visceral, 14, autosomal, 621080
CITED2	n.t.b					Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431
COL3A1	n.t.b					Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
COQ2	n.t.b					{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ7	n.t.b					Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
CORIN	n.t.b					?Cardiomyopathy, familial hypertrophic, 30, atrial, 620734;Preeclampsia/eclampsia 5, 614595
COX15	n.t.b					Mitochondrial complex IV deficiency, nuclear type 6, 615119

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CPT1A	n.t.b					CPT deficiency, hepatic, type IA, 255120
CPT2	n.t.b					{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	n.t.b					Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPTO	n.t.b					
CRYAB	n.t.b					Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
CSRP3	n.t.b					?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124
CTNNA3	n.t.b					Arrhythmogenic right ventricular dysplasia 13, 615616
CTNND1	n.t.b					Blepharocheilodontic syndrome 2, 617681

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DCHS1	n.t.b					Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DES	n.t.b					Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DMD	n.t.b					Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DNAH5	n.t.b					Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	n.t.b					Ciliary dyskinesia, primary, 40, 618300
DNAJC19	n.t.b					3-methylglutaconic aciduria, type V, 610198
DOCK6	n.t.b					Adams-Oliver syndrome 2, 614219
DOLK	n.t.b					Congenital disorder of glycosylation, type Im, 610768
DOT1L	n.t.b					Nil-Deshwar neurodevelopmental syndrome, 621265
DPM3	n.t.b					?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937

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DPP6	n.t.b					Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956
DSC2	n.t.b					Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	n.t.b					Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193
DSP	n.t.b					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	n.t.b					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	n.t.b					Robinow syndrome, autosomal dominant 3, 616894

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DYRK1A	n.t.b					Intellectual developmental disorder, autosomal dominant 7, 614104
DZIP1	n.t.b					Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
EEF1A2	n.t.b					Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EHMT1	n.t.b					Kleefstra syndrome 1, 610253
EIF2AK4	n.t.b					Pulmonary venoocclusive disease 2, 234810
EIF3A	n.t.b					
EIF3B	n.t.b					
ELAC2	n.t.b					{Prostate cancer, hereditary, 2}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
ELN	n.t.b					Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
EMD	n.t.b					Cardiomyopathy, dilated, 3C, 301163;Emery-Dreifuss muscular dystrophy 1, X-linked, 310300

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ENPP1	n.t.b					{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	n.t.b					
FAH	n.t.b					Tyrosinemia, type I, 276700
FBN1	n.t.b					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	n.t.b					Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBXO32	n.t.b					
FGF12	n.t.b					Developmental and epileptic encephalopathy 47, 617166
FGF8	n.t.b					Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

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FHL1	n.t.b					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	n.t.b					
FHOD3	n.t.b					Cardiomyopathy, familial hypertrophic, 28, 619402
FKRP	n.t.b					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

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FKTN	n.t.b					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	n.t.b					Cardiomyopathy, dilated, 2J, 620635
FLNA	n.t.b					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

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FLNC	n.t.b					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	n.t.b					Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FNIP1	n.t.b					Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXC2	n.t.b					Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXH1	n.t.b					
FOXJ1	n.t.b					Ciliary dyskinesia, primary, 43, 618699
GAA	n.t.b					Pompe disease, late-onset, 621314;Pompe disease, infantile-onset, 232300

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GATA4	n.t.b					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	n.t.b					Congenital heart defects, multiple types, 5, 617912
GATA6	n.t.b					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	n.t.b					?Cardiomyopathy, dilated, 2B, 614672
GATB	n.t.b					?Combined oxidative phosphorylation deficiency 41, 618838
GATC	n.t.b					Combined oxidative phosphorylation deficiency 42, 618839
GBE1	n.t.b					Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GDF1	n.t.b					Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GDF2	n.t.b					Telangiectasia, hereditary hemorrhagic, type 5, 615506

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GET3	n.t.b					?Cardiomyopathy, dilated, 2H, 620203
GJA5	n.t.b					Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770
GLA	n.t.b					Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLB1	n.t.b					GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLIS1	n.t.b					
GLYR1	n.t.b					
GMPPB	n.t.b					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	n.t.b					Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
GNPTAB	n.t.b					Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500

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GPD1L	n.t.b					Brugada syndrome 2, 611777
HADHA	n.t.b					HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	n.t.b					Mitochondrial trifunctional protein deficiency 2, 620300
HAND1	n.t.b					
HAND2	n.t.b					
HCN2	n.t.b					Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	n.t.b					
HCN4	n.t.b					Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
HECTD1	n.t.b					
HEY2	n.t.b					
HFE	n.t.b					Hemochromatosis, type 1, 235200
HJV	n.t.b					Hemochromatosis, type 2A, 602390
HSPB6	n.t.b					

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HSPD1	n.t.b					Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
IDUA	n.t.b					Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
ILK	n.t.b					
ISL1	n.t.b					
ITGA7	n.t.b					Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITPA	n.t.b					[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
JAG1	n.t.b					?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	n.t.b					Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873
JUP	n.t.b					Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
KANSL1	n.t.b					Koolen-De Vries syndrome, 610443

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KAT6B	n.t.b					SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KBTBD13	n.t.b					Nemaline myopathy 6, autosomal dominant, 609273
KCNA2	n.t.b					Developmental and epileptic encephalopathy 32, 616366
KCNA5	n.t.b					Atrial fibrillation, familial, 7, 612240
KCND2	n.t.b					
KCND3	n.t.b					Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNE1	n.t.b					Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNE2	n.t.b					Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493
KCNE3	n.t.b					?Brugada syndrome 6, 613119
KCNE4	n.t.b					
KCNE5	n.t.b					
KCNH2	n.t.b					Short QT syndrome 1, 609620;Long QT syndrome 2, 613688

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KCNJ11	n.t.b					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	n.t.b					Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNJ5	n.t.b					Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677
KCNJ8	n.t.b					
KCNK3	n.t.b					Developmental delay with sleep apnea, 621402;Pulmonary hypertension, primary, 4, 615344
KCNN3	n.t.b					Zimmermann-Laband syndrome 3, 618658
KCNQ1	n.t.b					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	n.t.b					{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089

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KIF20A	n.t.b					?Cardiomyopathy, familial restrictive, 6, 619433
KLF13	n.t.b					
KLHL24	n.t.b					Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294
KMT2A	n.t.b					Wiedemann-Steiner syndrome, 605130
KMT2D	n.t.b					Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920

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KRAS	n.t.b					Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;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;RAS-associated autoimmune leukoproliferative disorder 2, somatic, 621582;Bladder cancer, somatic, 109800
LAMA2	n.t.b					Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA4	n.t.b					Cardiomyopathy, dilated, 1JJ, 615235
LAMP2	n.t.b					Danon disease, 300257
LBX1	n.t.b					?Central hypoventilation syndrome, congenital, 3, 619483

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LDB3	n.t.b					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	n.t.b					Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500
LIMS2	n.t.b					?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LMCD1	n.t.b					
LMNA	n.t.b					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	n.t.b					Cardiomyopathy, dilated, 2G, 619897

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LOX	n.t.b					Aortic aneurysm, familial thoracic 10, 617168
LRRC10	n.t.b					
LTBP2	n.t.b					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	n.t.b					Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MED13L	n.t.b					Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MEGF8	n.t.b					Carpenter syndrome 2, 614976
MEIS2	n.t.b					Cleft palate, cardiac defects, and impaired intellectual development, 600987
MESP1	n.t.b					
MGRN1	n.t.b					
MIB1	n.t.b					Left ventricular noncompaction 7, 615092
MIPEP	n.t.b					Combined oxidative phosphorylation deficiency 31, 617228
MLYCD	n.t.b					Malonyl-CoA decarboxylase deficiency, 248360

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

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MYH7	n.t.b					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	n.t.b					
MYL2	n.t.b					Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	n.t.b					Cardiomyopathy, hypertrophic, 8, 608751
MYL4	n.t.b					?Atrial fibrillation, familial, 18, 617280
MYL7	n.t.b					
MYLK3	n.t.b					
MYO5B	n.t.b					Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868
MYO6	n.t.b					Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821

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MYOM1	n.t.b					
MYOT	n.t.b					Myopathy, myofibrillar, 3, 609200
MYOZ2	n.t.b					Cardiomyopathy, hypertrophic, 16, 613838
MYPN	n.t.b					Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248
MYRF	n.t.b					Nanophthalmos 1, 600165;Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
MYZAP	n.t.b					Cardiomyopathy, dilated, 2K, 620894
NAA15	n.t.b					Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NDUFAF1	n.t.b					Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFB11	n.t.b					Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NEBL	n.t.b					

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NEXN	n.t.b					Cardiomyopathy, dilated, 2M, autosomal recessive, 621261;Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876
NF1	n.t.b					Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NKX2-5	n.t.b					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	n.t.b					Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NODAL	n.t.b					Heterotaxy, visceral, 5, autosomal, 270100
NONO	n.t.b					Intellectual developmental disorder, X-linked syndromic 34, 300967
NOS1AP	n.t.b					Nephrotic syndrome, type 22, 619155

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NOTCH1	n.t.b					Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	n.t.b					Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NPHP4	n.t.b					Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NPPA	n.t.b					Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201
NPPB	n.t.b					
NR2F2	n.t.b					46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NRAP	n.t.b					Cardiomopathy, dilated, 2N, 621595
NSD1	n.t.b					Sotos syndrome, 117550
NUP155	n.t.b					?Atrial fibrillation 15, 615770
ODAD1	n.t.b					Ciliary dyskinesia, primary, 20, 615067
ODAD2	n.t.b					Ciliary dyskinesia, primary, 23, 615451
OLA1	n.t.b					
PBX1	n.t.b					Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	n.t.b					Propionicacidemia, 606054
PCCB	n.t.b					Propionicacidemia, 606054

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PDLIM3	n.t.b					
PDLIM5	n.t.b					
PEX5	n.t.b					Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	n.t.b					Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PGM1	n.t.b					Congenital disorder of glycosylation, type It, 614921
PHKA1	n.t.b					Muscle glycogenosis, 300559
PHYH	n.t.b					Refsum disease, 266500
PITX2	n.t.b					Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PKD1L1	n.t.b					Heterotaxy, visceral, 8, autosomal, 617205
PKP2	n.t.b					Arrhythmogenic right ventricular dysplasia 9, 609040
PLD1	n.t.b					Cardiac valvular dysplasia 1, 212093
PLEKHM2	n.t.b					

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PLN	n.t.b					Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874
PLXND1	n.t.b					Congenital heart defects, multiple types, 9, 620294
PMM2	n.t.b					Congenital disorder of glycosylation, type Ia, 212065
PNPLA2	n.t.b					Neutral lipid storage disease with myopathy, 610717
POMT1	n.t.b					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	n.t.b					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	n.t.b					Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812

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

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QRSL1	n.t.b					Combined oxidative phosphorylation deficiency 40, 618835
RAF1	n.t.b					Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RANGRF	n.t.b					
RBCK1	n.t.b					Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX2	n.t.b					
RBM20	n.t.b					Cardiomyopathy, dilated, 1DD, 613172
RIT1	n.t.b					Noonan syndrome 8, 615355
ROBO4	n.t.b					Aortic valve disease 3, 618496
RPL3L	n.t.b					Cardiomyopathy, dilated, 2D, 619371
RPS6KB1	n.t.b					
RRAD	n.t.b					
RRAGC	n.t.b					Long-Olsen-Distelmaier syndrome, 620609
RYR2	n.t.b					Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000
SCN10A	n.t.b					Episodic pain syndrome, familial, 2, 615551

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SCN1B	n.t.b					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	n.t.b					Atrial fibrillation, familial, 14, 615378
SCN3B	n.t.b					Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4B	n.t.b					Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN5A	n.t.b					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	n.t.b					Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377

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SDHA	n.t.b					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	n.t.b					Thyroid hormone metabolism, abnormal, 1, 609698
SF3B2	n.t.b					Craniofacial microsomia, 164210
SGCA	n.t.b					Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	n.t.b					Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	n.t.b					Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	n.t.b					Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SHMT2	n.t.b					Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC2	n.t.b					Noonan syndrome-like with loose anagen hair 1, 607721
SHOX2	n.t.b					
SHROOM3	n.t.b					

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

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SMARCA4	n.t.b					Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SNTA1	n.t.b					Long QT syndrome 12, 612955
SOD2	n.t.b					{Microvascular complications of diabetes 6}, 612634
SOS1	n.t.b					Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOX7	n.t.b					
SRF	n.t.b					
SRI	n.t.b					
STRA6	n.t.b					Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
SURF1	n.t.b					Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SVIL	n.t.b					Myofibrillar myopathy 10, 619040
TAB2	n.t.b					Congenital heart defects, nonsyndromic, 2, 614980
TAF1	n.t.b					Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TAF1A	n.t.b					
TAFAZZIN	n.t.b					Barth syndrome, 302060

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

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

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

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TTN	n.t.b					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	n.t.b					Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
TULP3	n.t.b					Hepatorenocardiac degenerative fibrosis, 619902
TXNRD2	n.t.b					?Glucocorticoid deficiency 5, 617825
UNC45B	n.t.b					?Cataract 43, 616279;Myofibrillar myopathy 11, 619178
VCL	n.t.b					Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255
VEZF1	n.t.b					?Cardiomyopathy, dilated, 1OO, 620247
XIRP2	n.t.b					
XK	n.t.b					McLeod syndrome, 300842
ZBTB17	n.t.b					

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ZFPM2	n.t.b					Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZIC3	n.t.b					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