Panel de Cardiología

Incluye los genes más relevantes para arritmias, cardiopatías congénitas y miocardiopatías. Los síndromes incluyeron: QT largo y corto, síndrome de Brugada, taquicardia ventricular polimórfica catecolaminérgica, miocardiopatías dilatadas e hipertróficas y defectos cardíacos congénitos. Además, este panel incluye anomalías vasculares, como dolicoectasia y telangiectasia hemorrágica hereditaria. El panel no incluye análisis de *PKD1*.

Síndromes y trastornos comunes cubiertos

Miocardiopatía arritmogénica del ventrículo derecho

Síndrome de Brugada

Taquicardia ventricular polimórfica catecolaminérgica

Defectos cardíacos congénitos

Miocardiopatía dilatada

Dolicoectasia

Síndromes de arritmia hereditaria

Telangiectasia hemorrágica hereditaria

Síndrome de heterotaxia

Miocardiopatía hipertrófica

Hipomagnesemia

Síndrome de QT largo

Síndrome de QT corto

Nº de genes:	323
Entrega:	25 días
Cobertura:	≥99,5% ≥20x Cobertura media con profundidad ≥ 150x
Detalles:	Análisis CNV incluido

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (gen)	Enfermedades asociadas (OMIM)	Herencia
ABCC9	601439	Hypertrichotic osteochondrodysplasia;Intellectual disability and myopathy syndrome;Cardiomyopathy, dilated, 1O; Atrial fibrillation, familial, 12	AD, AR
ACTA1	102610	Nemaline myopathy 3, AD or recessive;?Myopathy, scapulohumeroperoneal;Myopathy, congenital, with fiber-type disproportion 1;Myopathy, actin, congenital, with cores;Myopathy, actin, congenital, with excess of thin myofilaments	AD, AR, AD
ACTA2	102620	Aortic aneurysm, familial thoracic 6;Moyamoya disease 5;Multisystemic smooth muscle dysfunction syndrome	AD
ACTC1	102540	Atrial septal defect 5;Left ventricular noncompaction 4;Cardiomyopathy, hypertrophic, 11;Cardiomyopathy, dilated, 1R	AD
ACTN2	102573	Cardiomyopathy, dilated, 1AA, with or without LVNC;Myopathy, congenital with structured cores and Z-line abnormalities;Cardiomyopathy, hypertrophic, 23, with or without LVNC;Myopathy, distal, 6, adult onset	AD
ACVR2B	602730	Heterotaxy, visceral, 4, autosomal	

ACVRL1	601284	Telangiectasia, hereditary hemorrhagic, type 2	AD
ADAMTS10	608990	Weill-Marchesani syndrome 1, recessive	AR
ADAMTS19	607513		
AGL	610860	Glycogen storage disease IIIb;Glycogen storage disease IIIa	AR
AKAP9	604001	Long QT syndrome 11	AD
ALMS1	606844	Alstrom syndrome	AR
ALPK3	617608	Cardiomyopathy, familial hypertrophic 27	AR
ANK2	106410	Long QT syndrome 4;Cardiac arrhythmia, ankyrin-B-related	AD
ANKRD1	609599		
ANKS6	615370	Nephronophthisis 16	AR
ARHGAP31	610911	Adams-Oliver syndrome 1	AD
ATM	607585	Breast cancer, susceptibility to;Ataxia-telangiectasia	AD, SM, AR
B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	AR
BAG3	603883	Cardiomyopathy, dilated, 1HH;Myopathy, myofibrillar, 6	AD
BCOR	300485	Microphthalmia, syndromic 2	XLD

BMPR2	600799	Pulmonary hypertension, familial primary, 1, with or without HHT;Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated;Pulmonary venoocclusive disease 1	AD
BRAF	164757	Cardiofaciocutaneous syndrome;Adenocarcinoma of lung, somatic;Noonan syndrome 7;Colorectal cancer, somatic;Melanoma, malignant, somatic,;LEOPARD syndrome 3	AD
CACNA1C	114205	Long QT syndrome 8;Brugada syndrome 3;Timothy syndrome	AD
CACNA2D1	114204		
CACNB2	600003	Brugada syndrome 4	
CALM1	114180	Long QT syndrome 14;Ventricular tachycardia, catecholaminergic polymorphic, 4	AD
CALM2	114182	Long QT syndrome 15	AD
CALM3	114183	Ventricular tachycardia, catecholaminergic polymorphic 6;Long QT syndrome 16	AD
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2	AR
CAV3	601253	Rippling muscle disease 2;Cardiomyopathy, familial hypertrophic;Creatine phosphokinase, elevated serum;Long QT syndrome 9;Myopathy, distal, Tateyama type	AD, AD, DD
CAVIN4	617714		
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia;?Juvenile myelomonocytic leukemia	AD, AD, SM

CCDC103	614677	Ciliary dyskinesia, primary, 17	AR
CCDC39	613798	Ciliary dyskinesia, primary, 14	AR
CCDC40	613799	Ciliary dyskinesia, primary, 15	AR
CDH2	114020	Agenesis of corpus callosum, cardiac, ocular, and genital syndrome;Arrhythmogenic right ventricular dysplasia, familial, 14	AD
CFAP298	615494	Ciliary dyskinesia, primary, 26	AR
CFAP300	618058	Ciliary dyskinesia, primary, 38	AR
CFAP53	614759	Heterotaxy, visceral, 6, AR	AR
CHD7	608892	CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CITED2	602937	Atrial septal defect 8;Ventricular septal defect 2	AD
CLDN16	603959	Hypomagnesemia 3, renal	AR
CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement	AR
CNNM2	607803	Hypomagnesemia 6, renal;Hypomagnesemia, seizures, and mental retardation	AD, AD, AR
COL1A1	120150	Ehlers-Danlos syndrome, arthrochalasia type, 1;Bone mineral density variation QTL, osteoporosis;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type I;Caffey disease;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1;Osteogenesis imperfecta, type II	AD

COL1A2	120160	Osteoporosis, postmenopausal;Ehlers-Danlos syndrome, cardiac valvular type;Ehlers-Danlos syndrome, arthrochalasia type, 2;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2;Osteogenesis imperfecta, type II	AD, AR
COL3A1	120180	Ehlers-Danlos syndrome, vascular type;Polymicrogyria with or without vascular-type EDS	AD, AR
COL4A1	120130	?Retinal arteries, tortuosity of;Hemorrhage, intracerebral, susceptibility to;Microangiopathy and leukoencephalopathy, pontine, AD;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps;Brain small vessel disease with or without ocular anomalies	AD
COL4A2	120090	Hemorrhage, intracerebral, susceptibility to;Brain small vessel disease 2	AD
COL5A1	120215	Fibromuscular dysplasia, multifocal;Ehlers-Danlos syndrome, classic type, 1	AD
COL5A2	120190	Ehlers-Danlos syndrome, classic type, 2	AD
COX15	603646	Mi complex IV deficiency, nuclear type 6	AR
CPT2	600650	CPT II deficiency, myopathic, stress-induced;CPT II deficiency, infantile;Encephalopathy, acute, infection-induced, 4, susceptibility to;CPT II deficiency, lethal neonatal	AD, AR, AR
CREBBP	600140	Menke-Hennekam syndrome 1;Rubinstein-Taybi syndrome 1	AD
CRELD1	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome;Atrioventricular septal defect, susceptibility to, 2	AD

CRYAB	123590	Cataract 16, multiple types;Cardiomyopathy, dilated, 1II;Myopathy, myofibrillar, 2;Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related	AD, AR, AD, AR
CSRP3	600824	?Cardiomyopathy, dilated, 1M;Cardiomyopathy, hypertrophic, 12	AD
CTNNA3	607667	Arrhythmogenic right ventricular dysplasia, familial, 13	AD
DES	125660	Cardiomyopathy, dilated, 1I;Myopathy, myofibrillar, 1;Scapuloperoneal syndrome, neurogenic, Kaeser type	AD, AD, AR
DLL4	605185	Adams-Oliver syndrome 6	AD
DMD	300377	Cardiomyopathy, dilated, 3B;Duchenne muscular dystrophy;Becker muscular dystrophy	XL, XLR
DNAAF1	613190	Ciliary dyskinesia, primary, 13	AR
DNAAF2	612517	Ciliary dyskinesia, primary, 10	AR
DNAAF3	614566	Ciliary dyskinesia, primary, 2	AR
DNAAF4	608706	Dyslexia, susceptibility to, 1;Ciliary dyskinesia, primary, 25	AD, AR
DNAAF5	614864	Ciliary dyskinesia, primary, 18	AR
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus	AR
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus	AR
DNAH9	603330	Ciliary dyskinesia, primary, 40	AR
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus	AR

DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus	AR
DNAJC19	608977	3-methylglutaconic aciduria, type V	AR
DNAL1	610062	Ciliary dyskinesia, primary, 16	AR
DOLK	610746	Congenital disorder of glycosylation, type Im	AR
DPP6	126141	Ventricular fibrillation, paroxysmal familial, 2;Mental retardation, AD 33	AD
DSC2	125645	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair;Arrhythmogenic right ventricular dysplasia 11	AD, AR
DSG2	125671	Arrhythmogenic right ventricular dysplasia 10;Cardiomyopathy, dilated, 1BB	AD
DSP	125647	Cardiomyopathy, dilated, with woolly hair and keratoderma;Arrhythmogenic right ventricular dysplasia 8;Keratosis palmoplantaris striata II;Skin fragility-woolly hair syndrome;Epidermolysis bullosa, lethal acantholytic;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis	AR, AD
DTNA	601239	Left ventricular noncompaction 1, with or without congenital heart defects	AD
EFEMP2	604633	Cutis laxa, AR, type IB	AR
EGF	131530	Hypomagnesemia 4, renal	AR
EHMT1	607001	Kleefstra syndrome 1	AD
ELAC2	605367	Prostate cancer, hereditary, 2, susceptibility to;Combined oxidative phosphorylation deficiency 17	AR

ELN	130160	Cutis laxa, AD;Supravalvar aortic stenosis	AD
EMD	300384	Emery-Dreifuss muscular dystrophy 1, XL	XLR
ENG	131195	Telangiectasia, hereditary hemorrhagic, type 1	AD
EOGT	614789	Adams-Oliver syndrome 4	AR
EP300	602700	Rubinstein-Taybi syndrome 2;Colorectal cancer, somatic;Menke-Hennekam syndrome 2	AD
EVC	604831	Ellis-van Creveld syndrome;?Weyers acrofacial dysostosis	AR, AD
EVC2	607261	Ellis-van Creveld syndrome;Weyers acrofacial dysostosis	AR, AD
EYA4	603550	Deafness, AD 10;?Cardiomyopathy, dilated, 1J	AD
FBN1	134797	Marfan lipodystrophy syndrome;Geleophysic dysplasia 2;Acromicric dysplasia;Marfan syndrome;Weill-Marchesani syndrome 2, dominant;Stiff skin syndrome;MASS syndrome;Ectopia lentis, familial	AD
FBN2	612570	Contractural arachnodactyly, congenital;Macular degeneration, early-onset	AD
FHL1	300163	Reducing body myopathy, XL 1b, with late childhood or adult onset;Scapuloperoneal myopathy, XLD;?Uruguay faciocardiomusculoskeletal syndrome;Myopathy, XL, with postural muscle atrophy;Reducing body myopathy, XL 1a, severe, infantile or early childhood onset;Emery-Dreifuss muscular dystrophy 6, XL	XL, XLD, XLR
FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without mental	AR

		retardation), type B, 5;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	
FKTN	607440	Cardiomyopathy, dilated, 1X;Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	AR
FLNA	300017	Frontometaphyseal dysplasia 1;Heterotopia, periventricular, 1;Terminal osseous dysplasia;Congenital short bowel syndrome;Otopalatodigital syndrome, type II;Melnick-Needles syndrome;Cardiac valvular dysplasia, XL;Intestinal pseudoobstruction, neuronal;?FG syndrome 2;Otopalatodigital syndrome, type I	XLR, XLD, XL
FLNC	102565	Cardiomyopathy, familial restrictive 5;Myopathy, myofibrillar, 5;Myopathy, distal, 4;Cardiomyopathy, familial hypertrophic, 26	AD
FOXC1	601090	Anterior segment dysgenesis 3, multiple subtypes;Axenfeld-Rieger syndrome, type 3	AD
FOXF1	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins	AD
FOXH1	603621		
FOXJ1	602291	Ciliary dyskinesia, primary, 43	AD
FXYD2	601814	Hypomagnesemia 2, renal	AD
GAA	606800	Glycogen storage disease II	AR

GAS8	605178	Ciliary dyskinesia, primary, 33	AR
GATA4	600576	Atrioventricular septal defect 4;?Testicular anomalies with or without congenital heart disease;Ventricular septal defect 1;Tetralogy of Fallot;Atrial septal defect 2	AD
GATA5	611496	Congenital heart defects, multiple types, 5	AD, AR
GATA6	601656	Persistent truncus arteriosus;Atrioventricular septal defect 5;Tetralogy of Fallot;Atrial septal defect 9;Pancreatic agenesis and congenital heart defects	AD
GDF1	602880	Congenital heart defects, multiple types, 6;Right atrial isomerism (Ivemark)	AD, AR
GDF2	605120	Telangiectasia, hereditary hemorrhagic, type 5	AD
GJA1	121014	Oculodentodigital dysplasia, AR;Atrioventricular septal defect 3;Syndactyly, type III;Craniometaphyseal dysplasia, AR;Palmoplantar keratoderma with congenital alopecia;Oculodentodigital dysplasia;Hypoplastic left heart syndrome 1;Erythrokeratodermia variabilis et progressiva 3	AR, AD
GJA5	121013	Atrial fibrillation, familial, 11;Atrial standstill, digenic (GJA5/SCN5A)	AD
GLA	300644	Fabry disease;Fabry disease, cardiac variant	XL
GNB5	604447	Intellectual developmental disorder with cardiac arrhythmia;Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	AR
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic	XLR

GPD1L	611778	Brugada syndrome 2	
HADHA	600890	HELLP syndrome, maternal, of pregnancy;LCHAD deficiency;Fatty liver, acute, of pregnancy;Mi trifunctional protein deficiency	AR
HAND1	602406		
HCCS	300056	Linear skin defects with multiple congenital anomalies 1	XLD
HCN4	605206	Sick sinus syndrome 2;Brugada syndrome 8;Epilepsy, idiopathic generalized, susceptibility to, 18	AD
HFE	613609	Porphyria variegata, susceptibility to;Alzheimer disease, susceptibility to;Hemochromatosis;[Transferrin serum level QTL2];Porphyria cutanea tarda, susceptibility to;Microvascular complications of diabetes 7	AD, AR, AD, AR
HRAS	190020	Thyroid carcinoma, follicular, somatic;Spitz nevus or nevus spilus, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Bladder cancer, somatic;Costello syndrome;Nevus sebaceous or woolly hair nevus, somatic;Congenital myopathy with excess of muscle spindles	AD
HTRA1	602194	Macular degeneration, age-related, 7;CARASIL syndrome;Cerebral arteriopathy, AD, with subcortical infarcts and leukoencephalopathy, type 2;Macular degeneration, age-related, neovascular type	AR, AD
ILK	602366		
JAG1	601920	Alagille syndrome 1;Charcot-Marie-Tooth disease, axonal, type 2HH;?Deafness, congenital heart defects, and posterior embryotoxon;Tetralogy of Fallot	AD

JPH2	605267	Cardiomyopathy, hypertrophic, 17;Cardiomyopathy, dilated, 2E	AD, AR
JUP	173325	Arrhythmogenic right ventricular dysplasia 12;Naxos disease	AD, AR
KANSL1	612452	Koolen-De Vries syndrome	AD
KCNA1	176260	Episodic ataxia/myokymia syndrome	AD
KCNA5	176267	Atrial fibrillation, familial, 7	AD
KCND3	605411	Brugada syndrome 9;Spinocerebellar ataxia 19	AD
KCNE1	176261	Jervell and Lange-Nielsen syndrome 2;Long QT syndrome 5	AR, AD
KCNE2	603796	Atrial fibrillation, familial, 4;Long QT syndrome 6	AD
KCNE3	604433	?Brugada syndrome 6	
KCNE5	300328		
KCNH2	152427	Long QT syndrome 2;Short QT syndrome 1;Long QT syndrome 2, acquired, susceptibility to	AD
KCNJ2	600681	Short QT syndrome 3;Atrial fibrillation, familial, 9;Andersen syndrome	AD
KCNJ5	600734	Hyperaldosteronism, familial, type III;Long QT syndrome 13	AD
KCNJ8	600935		
KCNK3	603220	Pulmonary hypertension, primary, 4	AD

KCNQ1	607542	Long QT syndrome 1, acquired, susceptibility to;Jervell and Lange-Nielsen syndrome;Atrial fibrillation, familial, 3;Short QT syndrome 2;Long QT syndrome 1	AD, AR
KDM6A	300128	Kabuki syndrome 2	XLD
KLF10	601878		
KMT2D	602113	Kabuki syndrome 1	AD
KRAS	190070	Arteriovenous malformation of the brain, somatic;Gastric cancer, somatic;Oculoectodermal syndrome, somatic;RAS-associated autoimmune leukoproliferative disorder;Pancreatic carcinoma, somatic;Lung cancer, somatic;Cardiofaciocutaneous syndrome 2;Bladder cancer, somatic;Leukemia, acute myeloid, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Breast cancer, somatic;Noonan syndrome 3	AD
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient;Muscular dystrophy, limb-girdle, AR 23	AR
LAMA4	600133	Cardiomyopathy, dilated, 1JJ	AD
LAMP2	309060	Danon disease	XLD
LDB3	605906	Cardiomyopathy, hypertrophic, 24;Myopathy, myofibrillar, 4;Cardiomyopathy, dilated, 1C, with or without LVNC;Left ventricular noncompaction 3	AD
LDLR	606945	LDL cholesterol level QTL2;Hypercholesterolemia, familial, 1	AD, AR

LDLRAP1	605747	Hypercholesterolemia, familial, 4	AR
LEFTY2	601877		
LMNA	150330	Malouf syndrome;Emery-Dreifuss muscular dystrophy 3, AR;Emery-Dreifuss muscular dystrophy 2, AD;Hutchinson-Gilford progeria;Muscular dystrophy, congenital;Restrictive dermopathy, lethal;Lipodystrophy, familial partial, type 2;Charcot-Marie-Tooth disease, type 2B1;Mandibuloacral dysplasia;Cardiomyopathy, dilated, 1A;Heart-hand syndrome, Slovenian type	AD, AR
LRRC56	618227	Ciliary dyskinesia, primary, 39	AR
LZTR1	600574	Noonan syndrome 2;Schwannomatosis-2, susceptibility to;Noonan syndrome 10	AR, AD
MAP2K1	176872	Cardiofaciocutaneous syndrome 3;Melorheostosis, isolated, somatic mosaic	AD
MAP2K2	601263	Cardiofaciocutaneous syndrome 4	AD
MED12	300188	Opitz-Kaveggia syndrome;Lujan-Fryns syndrome;Ohdo syndrome, XL;Hardikar syndrome	XLR, XLD
MED13L	608771	Transposition of the great arteries, dextro-looped 1;Mental retardation and distinctive facial features with or without cardiac defects	AD
MEIS2	601740	Cleft palate, cardiac defects, and mental retardation	AD
MFAP5	601103	Aortic aneurysm, familial thoracic 9	AD
MGP	154870	Keutel syndrome	AR

MIB1	608677	Left ventricular noncompaction 7	AD
MMP15	602261		
MMP21	608416	Heterotaxy, visceral, 7, autosomal	AR
MMP3	185250	Coronary heart disease, susceptibility to, 6	
MRAS	608435	Noonan syndrome 11	AD
MT-ATP6			
MT-ATP8			
MT-CO1			
MT-CO2			
MT-CO3			
MT-CYB			
MT-ND1			
MT-ND2			
MT-ND3			
MT-ND4			
MT-ND4L			

MT-ND5			
MT-ND6			
MT-RNR1			
MT-RNR2			
MT-TA			
MT-TC			
MT-TD			
MT-TE			
MT-TF			
MT-TG			
MT-TH			
MT-TI			
MT-TK			
MT-TL1			
MT-TL2			
MT-TM			

MT-TN			
MT-TP			
MT-TQ			
MT-TR			
MT-TS1			
MT-TS2			
MT-TT			
MT-TV			
MT-TW			
MT-TY			
MYBPC3	600958	Cardiomyopathy, hypertrophic, 4;Cardiomyopathy, dilated, 1MM;Left ventricular noncompaction 10	AD, AR, AD
MYH11	160745	Aortic aneurysm, familial thoracic 4;Visceral myopathy 2;Megacystis-microcolon-intestinal hypoperistalsis syndrome 2	AD, AR
MYH6	160710	Atrial septal defect 3;Cardiomyopathy, hypertrophic, 14;Cardiomyopathy, dilated, 1EE;Sick sinus syndrome 3	AD
MYH7	160760	Myopathy, myosin storage, AR;Scapuloperoneal syndrome, myopathic type;Left ventricular noncompaction 5;Cardiomyopathy, dilated, 1S;Cardiomyopathy, hypertrophic,	AR, AD, AD, DD

		1;Myopathy, myosin storage, AD;Laing distal myopathy	
MYL2	160781	Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy;Cardiomyopathy, hypertrophic, 10	AR, AD
MYL3	160790	Cardiomyopathy, hypertrophic, 8	AD, AR
MYL4	160770	?Atrial fibrillation, familial, 18	AD
MYLK	600922	Aortic aneurysm, familial thoracic 7;Megacystis-microcolon-intestinal hypoperistalsis syndrome	AD, AR
MYLK2	606566	Cardiomyopathy, hypertrophic, 1, digenic	AD, DD
MYO6	600970	Deafness, AD 22, with hypertrophic cardiomyopathy;Deafness, AD 22;Deafness, AR 37	AD, AR
MYOM1	603508		
MYOZ2	605602	Cardiomyopathy, hypertrophic, 16	AD
MYPN	608517	Cardiomyopathy, hypertrophic, 22;Cardiomyopathy, dilated, 1KK;Cardiomyopathy, familial restrictive, 4;Nemaline myopathy 11, AR	AD, AR
NEBL	605491		
NEXN	613121	Cardiomyopathy, hypertrophic, 20;Cardiomyopathy, dilated, 1CC	AD
NF1	613113	Watson syndrome;Leukemia, juvenile myelomonocytic;Neurofibromatosis, type 1;Neurofibromatosis, familial spinal;Neurofibromatosis-Noonan syndrome	AD, AD, SM
NIPBL	608667	Cornelia de Lange syndrome 1	AD

NKX2-5	600584	Hypoplastic left heart syndrome 2;Hypothyroidism, congenital nongoitrous, 5;Atrial septal defect 7, with or without AV conduction defects;Conotruncal heart malformations, variable;Ventricular septal defect 3;Tetralogy of Fallot	AD
NKX2-6	611770	Persistent truncus arteriosus;Conotruncal heart malformations	
NME8	607421	Ciliary dyskinesia, primary, 6	AR
NODAL	601265	Heterotaxy, visceral, 5	AD
NOTCH1	190198	Adams-Oliver syndrome 5;Aortic valve disease 1	AD
NOTCH2	600275	Alagille syndrome 2;Hajdu-Cheney syndrome	AD
NOTCH3	600276	?Myofibromatosis, infantile 2;Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1;Lateral meningocele syndrome	AD
NPPA	108780	Atrial fibrillation, familial, 6;Atrial standstill 2	AD, AR
NR2F2	107773	46,XX sex reversal 5;Congenital heart defects, multiple types, 4	AD
NRAS	164790	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Colorectal cancer, somatic;Neurocutaneous melanosis, somatic;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic;Noonan syndrome 6;Melanocytic nevus syndrome, congenital, somatic;Thyroid carcinoma, follicular, somatic;Epidermal nevus, somatic	AD
NSD1	606681	Sotos syndrome 1	AD

ODAD4	617095	Ciliary dyskinesia, primary, 35	AR
PDLIM3	605889		
PKD1L1	609721	Heterotaxy, visceral, 8, autosomal	AR
PKD2	173910	Polycystic kidney disease 2	AD
PKP2	602861	Arrhythmogenic right ventricular dysplasia 9	AD
PLN	172405	Cardiomyopathy, dilated, 1P;Cardiomyopathy, hypertrophic, 18	AD
PPP1CB	600590	Noonan syndrome-like disorder with loose anagen hair 2	AD
PRDM16	605557	Cardiomyopathy, dilated, 1LL;Left ventricular noncompaction 8	AD
PRKAG2	602743	Glycogen storage disease of heart, lethal congenital;Cardiomyopathy, hypertrophic 6;Wolff-Parkinson-White syndrome	AD
PRKAR1A	188830	Myxoma, intracardiac;Pigmented nodular adrenocortical disease, primary, 1;Carney complex, type 1;Acrodysostosis 1, with or without hormone resistance	AD
PRKG1	176894	Aortic aneurysm, familial thoracic 8	AD
PSEN1	104311	Cardiomyopathy, dilated, 1U;Pick disease;Alzheimer disease, type 3;Alzheimer disease, type 3, with spastic paraparesis and apraxia;?Acne inversa, familial, 3;Dementia, frontotemporal;Alzheimer disease, type 3, with spastic paraparesis and unusual plaques	AD
PSEN2	600759	Alzheimer disease-4;Cardiomyopathy, dilated, 1V	AD

PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1	AD
RAF1	164760	Noonan syndrome 5;LEOPARD syndrome 2;Cardiomyopathy, dilated, 1NN	AD
RANGRF	607954		
RARB	180220	Microphthalmia, syndromic 12	AD, AR
RASA1	139150	Basal cell carcinoma, somatic;Capillary malformation-arteriovenous malformation 1	AD
RBM10	300080	TARP syndrome	XLR
RBM20	613171	Cardiomyopathy, dilated, 1DD	AD
RIT1	609591	Noonan syndrome 8	AD
ROBO4	607528	Aortic valve disease 8	AD
RYR1	180901	Malignant hyperthermia susceptibility 1;King-Denborough syndrome;Central core disease;Neuromuscular disease, congenital, with uniform type 1 fiber;Minicore myopathy with external ophthalmoplegia	AD, AD, AR, AR
RYR2	180902	Arrhythmogenic right ventricular dysplasia 2;Ventricular tachycardia, catecholaminergic polymorphic, 1;Ventricular arrythmias due to cardiac ryanodine receptor calcium release deficiency syndrome	AD
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome;Townes-Brocks syndrome 1	AD
SALL4	607343	IVIC syndrome;Duane-radial ray syndrome	AD

SCN10A	604427	Episodic pain syndrome, familial, 2	AD
SCN1B	600235	Atrial fibrillation, familial, 13;Epilepsy, generalized, with febrile seizures plus, type 1;Brugada syndrome 5;Cardiac conduction defect, nonspecific;Developmental and epileptic encephalopathy 52	AD, AR
SCN2B	601327	Atrial fibrillation, familial, 14	AD
SCN3B	608214	Brugada syndrome 7;Atrial fibrillation, familial, 16	AD
SCN4B	608256	Atrial fibrillation, familial, 17;Long QT syndrome 10	AD
SCN5A	600163	Heart block, nonprogressive;Ventricular fibrillation, familial, 1;Sick sinus syndrome 1;Brugada syndrome 1;Heart block, progressive, type IA;Atrial fibrillation, familial, 10;Long QT syndrome 3;Cardiomyopathy, dilated, 1E;Sudden infant death syndrome, susceptibility to	AD, AR
SCO2	604272	Myopia 6;Mi complex IV deficiency, nuclear type 2	AD, AR
SDHA	600857	Neurodegeneration with ataxia and late-onset optic atrophy;Cardiomyopathy, dilated, 1GG;Leigh syndrome;Mi respiratory chain complex II deficiency;Paragangliomas 5	AD, AR, AR, Mi
SELENON	606210	Muscular dystrophy, rigid spine, 1;Myopathy, congenital, with fiber-type disproportion	AR, AD, AR
SEMA3A	603961	Hypogonadotropic hypogonadism 16 with or without anosmia	AD
SGCD	601411	Muscular dystrophy, limb-girdle, AR 6;Cardiomyopathy, dilated, 1L	AR
SGCG	608896	Muscular dystrophy, limb-girdle, AR 5	AR

SHOC2	602775	Noonan syndrome-like with loose anagen hair 1	AD
SKI	164780	Shprintzen-Goldberg syndrome	AD
SLC12A3	600968	Gitelman syndrome	AR
SLC22A5	603377	Carnitine deficiency, systemic primary	AR
SLC25A4	103220	Mi DNA depletion syndrome 12B (cardiomyopathic type) AR;Mi DNA depletion syndrome 12A (cardiomyopathic type) AD;Progressive external ophthalmoplegia with Mi DNA deletions, AD 2	AR, AD
SLC2A10	606145	Arterial tortuosity syndrome	AR
SLMAP	602701		
SMAD3	603109	Loeys-Dietz syndrome 3	AD
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Pancreatic cancer, somatic;Myhre syndrome;Polyposis, juvenile intestinal	AD
SMAD6	602931	Craniosynostosis 7, susceptibility to;Aortic valve disease 2;Radioulnar synostosis, nonsyndromic	AD
SMC3	606062	Cornelia de Lange syndrome 3	AD
SNTA1	601017	Long QT syndrome 12	AD
SOS1	182530	Noonan syndrome 4;?Fibromatosis, gingival, 1	AD

SOS2	601247	Noonan syndrome 9	AD
SOX2	184429	Microphthalmia, syndromic 3;Optic nerve hypoplasia and abnormalities of the central nervous system	AD
SPAG1	603395	Ciliary dyskinesia, primary, 28	AR
STRA6	610745	Microphthalmia, isolated, with coloboma 8;Microphthalmia, syndromic 9	AR
SYNE1	608441	Arthrogryposis multiplex congenita 3, myogenic type;Emery-Dreifuss muscular dystrophy 4, AD;Spinocerebellar ataxia, AR 8	AR, AD
SYNE2	608442	Emery-Dreifuss muscular dystrophy 5, AD	AD
TAB2	605101	Congenital heart defects, nonsyndromic, 2	AD
TAFAZZIN	300394	Barth syndrome	XLR
TBX1	602054	Velocardiofacial syndrome;Tetralogy of Fallot;DiGeorge syndrome;Conotruncal anomaly face syndrome	AD
TBX20	606061	Atrial septal defect 4	
TBX3	601621	Ulnar-mammary syndrome	AD
TBX5	601620	Holt-Oram syndrome	AD
TCAP	604488	Muscular dystrophy, limb-girdle, AR 7;Cardiomyopathy, hypertrophic, 25	AR, AD
TECRL	617242	Ventricular tachycardia, catecholaminergic polymorphic, 3	AR
TFAP2B	601601	Patent ductus arteriosus 2;Char syndrome	AD

TGFB2	190220	Loeys-Dietz syndrome 4	AD
TGFB3	190230	Loeys-Dietz syndrome 5;Arrhythmogenic right ventricular dysplasia 1	AD
TGFBR1	190181	Loeys-Dietz syndrome 1;Multiple self-healing squamous epithelioma, susceptibility to	AD
TGFBR2	190182	Colorectal cancer, hereditary nonpolyposis, type 6;Loeys-Dietz syndrome 2;Esophageal cancer, somatic	AD
TLL1	606742	Atrial septal defect 6	AD
TMEM43	612048	Arrhythmogenic right ventricular dysplasia 5;Emery-Dreifuss muscular dystrophy 7, AD	AD
TNNC1	191040	Cardiomyopathy, dilated, 1Z;Cardiomyopathy, hypertrophic, 13	AD
TNNI3	191044	Cardiomyopathy, hypertrophic, 7;Cardiomyopathy, familial restrictive, 1;Cardiomyopathy, dilated, 1FF;?Cardiomyopathy, dilated, 2A	AD, AR
TNNI3K	613932	Cardiac conduction disease with or without dilated cardiomyopathy	AD
TNNT2	191045	Left ventricular noncompaction 6;Cardiomyopathy, familial restrictive, 3;Cardiomyopathy, dilated, 1D;Cardiomyopathy, hypertrophic, 2	AD
TPM1	191010	Cardiomyopathy, dilated, 1Y;Left ventricular noncompaction 9;Cardiomyopathy, hypertrophic, 3	AD
TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness	AR
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive;Chilblain lupus;Vasculopathy, retinal, with cerebral leukodystrophy;Systemic lupus erythematosus, susceptibility to	AD, AR, AD

TRIM63	606131		
TRPM4	606936	Erythrokeratodermia veriabilis et progressiva 6;Progressive familial heart block, type IB	AD
TRPM6	607009	Hypomagnesemia 1, intestinal	AR
TTC12	610732	Ciliary dyskinesia, primary, 45	AR
TTN	188840	Cardiomyopathy, dilated, 1G;Muscular dystrophy, limb-girdle, AR 10;Tibial muscular dystrophy, tardive;Myopathy, myofibrillar, 9, with early respiratory failure;Salih myopathy;Cardiomyopathy, familial hypertrophic, 9	AR, AD
TTR	176300	[Dystransthyretinemic hyperthyroxinemia];Carpal tunnel syndrome, familial;Amyloidosis, hereditary, transthyretin-related	AD
VCL	193065	Cardiomyopathy, dilated, 1W;Cardiomyopathy, hypertrophic, 15	AD
ZEB2	605802	Mowat-Wilson syndrome	AD
ZFPM2	603693	Tetralogy of Fallot;Diaphragmatic hernia 3;46XY sex reversal 9	AD
ZIC3	300265	VACTERL association, XL;Congenital heart defects, nonsyndromic, 1, XL;Heterotaxy, visceral, 1, XL	XLR
ZMYND10	607070	Ciliary dyskinesia, primary, 22	AR