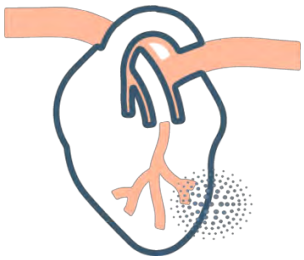
Comprehensive Cardiology Precision Panel



Overview

Cardiomyopathy is a group of conditions with a strong genetic background that structurally hinder the heart to pump out blood to the rest of the body due to weakness in the heart muscles. These diseases affect individuals of all ages and can lead to heart failure and sudden cardiac death. If there is a family history of cardiomyopathy it is strongly recommended to undergo genetic testing to be aware of the family risk, personal risk, and treatment options. Most types of cardiomyopathies are inherited in a dominant manner, which means that one altered copy of the gene is enough for the disease to present in an individual. The symptoms of cardiomyopathy are variable, and these diseases can present in different ways. There are 5 types of cardiomyopathies, the most common being hypertrophic cardiomyopathy:

1. Hypertrophic cardiomyopathy (HCM)
2. Dilated cardiomyopathy (DCM)
3. Restrictive cardiomyopathy (RCM)
4. Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)
5. Isolated Left Ventricular Non-Compaction Cardiomyopathy (LVNC).

Cardiac channelopathies are a group of inherited conditions that are associated with a defect in the cardiac ion channel function. These problems cause an increased susceptibility to abnormal heart rhythm (dysrhythmia), most often ventricular tachycardia or ventricular fibrillation that ultimately leads to sudden cardiac death (SCD). The differential diagnosis between ion channel disease and cardiomyopathies can be challenging on occasion as severe ventricular dysrhythmias can manifest in patients with cardiopathies or with structurally normal hearts.

The Igenomix Comprehensive Cardiology Precision Panel provides a comprehensive analysis of the most common mutations causing channelopathies, cardiomyopathies and sudden cardiac death using next-generation sequencing (NGS).

Indications

The Igenomix Comprehensive Cardiology Precision Panel is indicated in those cases where there is:

- Shortness of breath
- Fatigue
- Arrythmia (abnormal heart rhythm)
- Family history of arrhythmia
- Abnormal scans

- Ventricular tachycardia
- Ventricular fibrillation
- Chest Pain
- Dizziness
- Sudden cardiac death in the family

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis of a patient with personal or family history of cardiomyopathy, channelopathy or sudden cardiac death
- Early initiation of treatment with a multidisciplinary team for appropriate preventive ICD placement, pacemaker, pharmacologic therapy, or interventional procedures.
- Prognostic information and genetic counselling for family at risk.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

High Risk	Well studied Greater than 4-fold risk of developing one or more cancers Can cause a moderate risk for other cancers Guidelines or expert opinion for cancer screening and prevention
Moderate Risk	Well-studied 2- to 4-fold risk of developing one or more cancers May increase risk for other cancers Limited guidelines for screening and prevention
Research	Not as well-studied Precise lifetime risks and tumor spectrum not yet determined Guidelines for screening and prevention are limited or not available

GENE	RISK	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AARS2		Combined Oxidative Phosphorylation Deficiency 8, Progressive Leukoencephalopathy With Ovarian Failure	AR	100%	54 of 54
ABCC6		Generalized Arterial Calcification of Infancy, Pseudoxanthoma Elasticum	AD,AR	99%	346 of 349
ABCC9		Acromegaloid Facial Appearance Syndrome, Familial Atrial Fibrillation, Brugada Syndrome, Dilated Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy, Hypertrichosis-Acromegaloid Facial Appearance Syndrome, Hypertrichotic Osteochondrodysplasia	AD	100%	51 of 51
ACAD9		Acyl-CoA Dehydrogenase 9 Deficiency	AR	100%	62 of 62
ACADVL		Very Long Chain Acyl-CoA Dehydrogenase Deficiency	AR	100%	329 of 329
ACTA1		Childhood-Onset Nemaline Myopathy, Congenital Fiber-Type Disproportion Myopathy, Intermediate Nemaline Myopathy	AD,AR	100%	224 of 224
ACTA2		Familial Aortic Aneurysm, Familial Thoracic Aortic Aneurysm and Aortic Dissection, Moyamoya Disease , Multisystemic Smooth Muscle Dysfunction Syndrome	AD	100%	88 of 88
ACTC1	High Risk	Atrial Septal Defect Ostium Secundum, Dilated Cardiomyopathy, LeftVentricular Noncompaction, Familial Hypertrophic Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy	AD	99.93%	72 of 74
ACTN2		Dilated Cardiomyopathy With Or Without Left Ventricular Noncompaction, Familial Isolated Dilated Cardiomyopathy, Congenital Myopathy With Structured Cores And Z-line Abnormalities	AD	100%	56 of 56
AGK		Autosomal Recessive Congenital Cataract And Cardiomyopathy	AR	99.98%	33 of 33
AGL		Glycogen Storage Disease Due To Glycogen Debranching Enzyme Deficiency (type 3)	AR	100%	253 of 253
AGPAT2		Berardinelli-Seip Congenital Lipodystrophy	AR	100%	42 of 43
AKAP9		Brugada Syndrome, Long Qt Syndrome, Romano-Ward Syndrome	AD	98.34%	43 of 46
ALMS1		Alstrom Syndrome	AR	99.92%	302 of 305
ALPK3		Familial Hypertrophic Cardiomyopathy	AR	97.29%	7 of 7
ANK2		Ankyrin-B Related Cardiac Arrhythmia, Romano-Ward Syndrome	AD	99.98%	130 of 130
ANO5		Anoctamin-5-related Limb-Girdle Muscular Dystrophy, Anoctaminopathy, Gnathodiaphyseal Dysplasia, Miyoshi Muscular	AD,AR	99.78%	171 of 173
APOA1		Familial Visceral Amyloidosis, Apolipoprotein A-1 Deficiency	AD	99.89%	68 of 70

ATP6		Familial Infantile Bilateral Striatal Necrosis, Leber Hereditary Optic Neuropathy, Mitochondrial Dna-Associated Leigh Syndrome, MT-ATP6-Related Mitochondrial Spastic Paraplegia, Narp Syndrome, Retinitis Pigmentosa	MI	na	na
ATP8		Kearns-Sayre Syndrome		98.02%	NA of NA
ATPAF2		Nuclear-encoded ATPase Deficiency	AR	100%	2 of 2
BAG3	High Risk	Dilated Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy, Myofibrillar Myopathy	AD	100%	83 of 85
BRAF		Cardiofaciocutaneous Syndrome, Leopard Syndrome, Noonan Syndrome With Multiple Lentigines	AD	100%	80 of 80
CACNA1C	High Risk	Brugada Syndrome, Romano-Ward Syndrome, Timothy Syndrome	AD	99.80%	85 of 85
CACNB2		Brugada Syndrome	AD	99.84%	32 of 34
CALM1	High Risk	Catecholaminergic Polymorphic Ventricular Tachycardia, Long QT Syndrome, Romano-Ward Syndrome, Catecholaminergic PolymorphicVentricular Tachycardia	AD	100%	12 of 12
CALM2	High Risk	Catecholaminergic Polymorphic Ventricular Tachycardia, Long Qt Syndrome, Romano-Ward Syndrome	AD	98.71%	11 of 11
CALM3	High Risk	Catecholaminergic Polymorphic Ventricular Tachycardia, Long Qt Syndrome, Romano-Ward Syndrome	AD	100%	5 of 5
CALR3		Familial Isolated Hypertrophic Cardiomyopatht		100%	5 of 5
CAPN3		Calpain-3-related Limb-girdle Muscular Dystrophy	AD,AR	100%	503 of 505
CASQ2	High Risk	Catecholaminergic Polymorphic Ventricular Tachycardia With Or Without Atrial Dysfunction And/Or Dilated Cardiomyopathy	AD,AR	100%	39 of 40
CASZ1		Dopa Responsive Dystonoia		90.50%	6 of 6
CAV3		Familial Hypertrophic Cardiomyopathy, Elevated Serum Creatine Phosphokinase, Tateyama Type Distal Myopathy, Long Qt Syndrome, Rippling Muscle Disease, Rromano-Ward Syndrome	AD	100%	50 of 50
CBL		Aggressive Systemic Mastocytosis, Juvenile Myelomonocytic Leukemia, Noonan Syndrome	AD	100%	46 of 47
CDH2		Agenesis Of Corpus Callosum, Cardiac, Ocular, And Genital Syndrome, Familial Arrhythmogenic Right Ventricular Dysplasia	AD	99.98%	16 of 16
CHRM2		Dilated Cardiomyopathy		99.98%	1 of 1
COX15		Fatal Infantile Cardioencephalomyopathy Due To Cytochrome C Oxidasedeficiency 2, Leigh Syndrome With Leukodystrophy	AR,MI	100%	5 of 5
COX3		Leber Hereditary Optic Neuropathy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like episodes (MELAS)	MI	na	na
CPT2		Carnitine Palmitoyl Transferase II Deficiency, Carnitine Palmitoyl Transferase II Deficiency	AD,AR	99.99%	116 of 116
CRPPA		Congenital Muscular Dystrophy Without Intellectual Disability, ISPD-related Limb-girdle Muscular Dystrophy, Muscular Dystrophy-dystroglycanopathy (Congenital With Brain And Eye Anomalies), Type A, and Type C, Walker-Warburg Syndrome	AR	97.69%	NA of NA
CRYAB		Alpha-b Crystallin-Related Late-Onset Myopathy,alpha-b Crystallinopathy,cardiomyopathy, Dilated, 1ii; Cmd1ii,cataract, Posterior Polar, 2; Ctpp2cataract, Congenital Lamellar, Included,familial Isolated Dilated Cardiomyopathy,myopathy, Myofibrillar, Fatal Infantile Hypertonic, Alpha-b Crystallin-related	AD,AR	100%	30 of 30
CSRP3		Dilated Cardiomyopathy, Familial Hypertrophic, Familial Isolated Dilated Cardiomyopathy	AD	100%	36 of 36
CTNNA3		Familial Arrhythmogenic Right Ventricular Dysplasia	AD	99.97%	14 of 17
CYTB		Histiocytoid Cardiomyopathy, Leber Hereditary Optic Neuropathy, Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-likeepisodes (MELAS)	MI	98.80%	NA of NA
DBH		Congenital Dopamine Beta-hydroxylase Deficiency	AR	100%	11 of 11
DES	High Risk	Dilated Cardiomyopathy, Desminopathy, Familial Isolated Dilated Cardiomyopathy, Myofibrillar Myopathy, Neurogenic Scapuloperoneal Syndrome Kaeser Type	AD,AR	99.97%	133 of 134
DMD	High Risk	Becker Muscular Dystrophy, Duchenne Muscular Dystrophy, Familial Isolated Dilated Cardiomyopathy, Becker Type Muscular Dystrophy, X-linked Non-syndromic Intellectual Disability	X,XR,G	99.96%	NA of NA
DNAJC19		3-a Methylglutaconic Aciduria Type V, Dilated Cardiomyopathy With Ataxia	AR	100%	6 of 6
DOLK		Congenital Disorder Of Glycosylation, Type Im, Familial Isolated Dilated Cardiomyopathy	AR	99.98%	13 of 13
DPM3		Congenital Disorder Of Glycosylation, Type Io, Muscular Dystrophy-dystroglycanopathy Type B	AR	100%	4 of 4
DSC2	High Risk	Familial Arrhythmogenic Right Ventricular Dysplasia	AD,AR	100%	123 of 124
DSG2	High Risk	Famililal Arrhythmogenic Right Ventricular Dysplasia, Dilated Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy	AD	99.38%	167 of 169
DSP	High Risk	Familial Arrhythmogenic Right Ventricular Dysplasia, Dilated Cardiomyopathy With Woolly Hair And Keratoderma, Carvajal Syndrome, Lethal Acantholytic Epidermolysis Bullosa, Idiopathic Pulmonary Fibrosis, Keratosis Palmoplantaris Striata	AD,AR	99.91%	366 of 369
DTNA		Left Ventricular Noncompaction	AD	97%	10 of 10
DYSF		Distal Myopathy With Anterior Tibial Onset, Dysferlin-related, Limb-girdle Muscular Dystrophy, Miyoshi Myopathy	AR	100%	604 of 606
EEF1A2		Early Infantile Epileptic Encephalopathy, Autosomal Dominant Mental Retardation, Undetermined Early-onset Epileptic Encephalopathy	AD	100%	14 of 14
ELAC2		Combined Oxidative Phosphorylation Deficiency	AR	100%	32 of 32
EMD	High Risk	X-linked Emery-Dreifuss Muscular Dystrophy	X,XR,G	99.92%	NA of NA

ENPP1		Generalized Arterial Calcification of Infancy, Autosomal Recessive Hypophosphatemic Rickets, Cole Disease, Non-insulin Dependent Diabetes Mellitus, Pseudoxanthoma Elasticum	AD,AR,MU,P	96.59%	73 of 75
EPG5		Immunodeficiency With Cleft Lip/Palate, Cataract, Hypopigmentation,and Absent Corpus Callosum,Vvici Syndrome	AR	98.98%	73 of 73
ETFA		Multiple Acyl-CoA Dehydrogenase Deficiency	AR	92.33%	32 of 32
ETFB		Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100%	21 of 21
ETFDH		Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100%	221 of 222
FAH		Tyrosinemia Type 1	AR	100%	107 of 108
FBXL4		Mitochondrial DNA Depletion Syndrome	AR	99.26%	46 of 51
FBXO32		Skeletal Muscle Cancer		100%	2 of 2
FHL1	High Risk	X-linked Reducing Body Myopathy, X-linked Dominant Scapuloperoneal Myopathy, X-linked Faciocardiomusculoskeletal Syndrome, Emery-Dreifuss Muscular Dystrophy	X,XR,XD,G	99.98%	NA of NA
FHOD3	High Risk	Hypertrophic Cardiomyopathy, Hemochromatosis Type 2B		99.95%	35 of 35
FKRP		Congenital Muscular Dystrophy With Cerebellar Involvement and Intellectual Dissability, Limb-girdle Muscular Dystrophy, Muscle-eye-brain Disease, Muscular Dystrophy-dystroglycanopathy Type A and Type C, Walker-Warburg Syndrome	AR	99.90%	157 of 157
FKTN		Dilated Cardiomyopathy, Congenital Muscular Dystrophy Without Intellectual Disability, Fukuyama Congenital Muscular Dystrophy, Familial Isolated Dilated Cardiomyopathy, Muscle-eye-brain Disease, Muscular Dystrophy-dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A and Type B, Limb-girdle Muscular Dystrophy Type 2m, Walker-Warburg Syndrome	AR	98%	54 of 56
FLNC	High Risk	Familial Hypertrophic Cardiomyopathy, Distal Myopathy With Posterior Leg And Anterior Hand Involvement, Familial Isolated Restrictive Cardiomyopathy, Autosomal Dominant Filaminopathy	AD	100%	185 of 186
FOXD4		Chromosome 9P Deletion Syndrome, Dilated Cardiomyopathy		100%	1 of 1
FOXRED1		Isolated Complex I Deficiency, Leigh Syndrome With Leukodystrophy, Mitochondrial Complex I Deficiency, Nuclear Type	AR	100%	13 of 13
FXN		Friedreich Ataxia	AR	99.93%	52 of 52
GAA		Glycogen Storage Disease II	AR	100%	623 of 624
GATA4		46,XY Partial Gonadal Dysgenesis, Microdeletion Syndrome, Atrial Septal Defect Ostium Secundum, Atrioventricular Septal Defect, Partial Atrioventricular Septal Defect, Testicular Anomalies With Or Without Congenital Heart Disease, Tetralogy Of Fallot, Ventricular Septal Defect	AD	94.69%	108 of 130
GATA5		Multiple Types Congenital Heart Defects, Familial Bicuspid Aortic Valve, Tetralogy Of Fallot	AD,AR	87.02%	26 of 32
GATA6		Atrial Septal Ostium Secundum Type, Atrioventricular Septal Defect, Congenital Diaphragmatic Hernia, Conotruncal Heart Malformations, Truncus Arteriosus, Pancreatic Hypoplasia-Diabetes-Congenital Heart Disease Syndrome, Partial Atrioventricular Septal Defect, Tetralogy Of Fallot	AD,AR	84.19%	66 of 84
GATAD1		Dilated Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy	AR	88.20%	1 of 1
GATC		Combined Oxidative Phosphorylation Deficiency	AR	100%	1 of 1
GBE1		Adult Polyglucosan Body Disease, Glycogen Storage Disease IV, Adult Form Polyglucosan Body Disease	AR	99.95%	71 of 74
GFM1		Combined Oxidative Phosphorylation Deficiency	AR	100%	27 of 27
GLA	High Risk	Fabry Disease	X,XR,G	98%	NA of NA
GLB1		Type 1 and Type 2 GM1-gangliosidosis,Type 3 Morquio Syndrome	AR	100%	242 of 243
GMPPB		Congenital Muscular Dystrophy With Cerebellar Involvement and Intellectual Dissability, Congenital Myasthenic Syndromes With Glycosylation Defect, GMPPB-related Limb-girdle Muscular Dystrophy, Muscle-eye-brain Disease, Muscular Dystrophy-dystroglycanopathy (Congenital With Brain And Eyeanomalies) Type A, Type B and Type C	AR	99.95%	53 of 53
GSK3B		Alzheimer Disease, Diabetes Mellitus		99.91%	1 of 1
GTPBP3		Combined Oxidative Phosphorylation Defect Type 23	AR	99.94%	17 of 17
GUSB		Mucopolysaccharidosis Type 7	AR	100%	65 of 66
HADHA		Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency, Mitochondrial Trifunctional Protein Deficiency	AR	100%	75 of 75
HAND1		Multiple Congenital Cardiac Malformations		99.89%	9 of 9
HAND2		Familial Isolated Dilated Cardiomyopathy		99.19%	5 of 6
HCN4	High Risk	Brugada Syndrome, Sick Sinus Syndrome	AD	98.01%	40 of 41
HFE		Alzheimer Disease, Hemochromatosis, Porphyria Cutanea Tarda, Porphyria Variegata	AD,AR	100%	55 of 57
HRAS		Bladder Cancer, Costello Syndrome, Epidermal Nevus, Somatic Giant Pigmented Hairy Nevus, Linear Nevus Sebaceus Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Nonmedullary Thyroid Cancer	AD	100%	34 of 34
IDUA		Hurler Syndrome, Hurler-Scheie Syndrome	AR	99.73%	287 of 292
ILK		Focal Segmental Glomerulosclerosis and Dilated Cardiomyopathy		100%	14 of 14
JPH2		Familila Hypertrophic Cardiomyopath	AD	98.24%	17 of 17
JUP	High Risk	Famililal Arrhythmogenic Right Ventricular Dysplasia, Lethal Acantholytic Epidermolysis Bullosa, Naxos Disease	AD,AR	100%	56 of 56
KCNA5		Familial Atrial Fibrillation	AD	99.99%	33 of 33
KCNE1	High Risk	Jervell And Lange-Nielsen Syndrome, Long Qt Syndrome, Romano-Ward Syndrome	AD,AR	100%	53 of 53
KCNE2	High Risk	Familial Atrial Fibrillation, Long Qt Syndrome, Romano-Ward Syndrome	AD	100%	23 of 24
KCNH2	High Risk	Familial Short QT Syndrome, Long Qt Syndrome, Romano-Ward Syndrome	AD	98.69%	908 of 930
KCNJ2	High Risk	Andersen Cardiodysrhythmic Periodic Paralysis, Familial Atrial Fibrillation, Familial Short QT Syndrome	AD	100%	93 of 93

KCNJ5		Familial Hyperaldosteronism Type III, Long Qt Syndrome, Romano-Ward Syndrome	AD	99.52%	21 of 21
KCNQ1	High Risk	Familial Atrial Fibrillation, Beckwith-Wiedemann Syndrome, Familial Short Qt Syndrome, Jervell And Lange-nielsen Syndrome, Long Qt Syndrome, Romano-Ward Syndrome	AD,AR	93.23%	600 of 624
KLHL24		Generalized Epidermolysis Bullosa Simplex With Scarring And Hair Loss	AD	99.96%	8 of 8
KRAS		Aplasia Cutis Congenita With Epibulbar Dermoids, Arteriovenous Malformation Of The Brain, Somatic,bladder Cancer, Breast Cancer, Cardiofaciocutaneous Syndrome, Encephalocraniocutaneous Lipomatosis, Familial Pancreatic Carcinoma, Gastric Cancer, Acute Myeloids Leukemia, Linear Nevus Sebaceus Syndrome, Lung Cancer, Lynch Syndrome, Noonan Syndrome, RAS-associated Autoimmune Lymphoproliferative Syndrome Type IV, Schimmelpenning-Feuerstein-Mims Syndrome; Toriello-Lacassie-Droste Syndrome	AD	100%	38 of 38
LAMA2		Laminin Subunit Alpha 2-related Congenital Muscular Dystrophy, Congenital Merosin-Deficient 1A, Limb-girdle Muscular Dystrophy	AR	100%	363 of 377
LAMP2	High Risk	Danon Disease, Glycogen Storage Disease Due To Lamp-2 Deficiency	X,XD,G	99.96%	NA of NA
LARGE1		Congenital Muscular Dystrophy With Intellectual Disability, Muscle-Eye-Brain Disease, Muscular Dystrophy-dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Walker-Warburg Syndrome	AR	100%	NA of NA
LDB3		Dilated Cardiomyopathy With Or Without Left Ventricular Noncompaction, Left Ventricular Noncompaction, Markesbery-Griggs Type, Myofibrillar Myopathy	AD	100%	60 of 60
LEMD2		Congenital Cataract	AR	93.48%	3 of 3
LMNA	High Risk	Atypical Werner Syndrome, Emery-Dreifuss Muscular Dystrophy, Autosomal Semi-Dominant Severe Lipodystrophic Laminopathy, Dilated Cardiomyopathy, Charcot-Marie-Tooth Disease, Axonal, Type 2B1, Congenital Muscular Dystrophy Due To LMNA Mutation, Hutchinson-Gilford Progeria Syndrome, Malouf Syndrome, Mandibuloacral Dysplasia	AD,AR	100%	619 of 620
LMOD2		Familial Hypertrophic Cardiomyopathy, Megacystis-Microcolon-Intestinal Hypoperistalsis SYndrome		99.37%	1 of 1
LRRC10		Dilated Cardiomyopathy, Anomalous Left Coronary Artery from Pulmonary Artery		100%	5 of 5
LZTR1		Noonan Syndrome, Schwannomatosis	AD	99.99%	136 of 136
MAP2K1		Cardiofaciocutaneous Syndrome, Isolated Melorheostosis, Noonan Syndrome	AD	100%	31 of 31
MAP2K2		Cardiofaciocutaneous Syndrome, Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome	AD	100%	37 of 37
MAP3K8		Lung Cancer	AD	99.91%	1 of 1
MIPEP		Combined Oxidative Phosphorylation Deficiency	AR	99.84%	7 of 8
MLYCD		Malonyl-CoA Decarboxylase Deficiency	AR	93.84%	32 of 40
MRPL3		Combined Oxidative Phosphorylation Deficiency	AR	99.96%	4 of 4
MRPL44		Combined Oxidative Phosphorylation Deficiency	AR	99.75%	2 of 2
MRPS22		46,XX Gonadal Dysgenesis, Combined Oxidative Phosphorylation Deficiency, Ovarian Dysgenesis	AR	100%	10 of 10
MT-CO1		Myoglobinuria, Autosomal Recessive Pyridoxine-Refractory Sideroblastic Anemia		97.64%	NA of NA
MT-CO2		Mitochondrial Complex IV Deficiency		99.19%	NA of NA
MT-ND1		Leber Hereditar Optic Neuropathy		98.80%	NA of NA
MTO1		Combined Oxidative Phosphorylation Deficiency	AR	99.83%	31 of 31
MYBPC3	High Risk	Familial Hypertrophic Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy, Left Ventricular Noncompaction	AD,AR	99.95%	1072 of 1079
MYBPHL		Dilated Cardiomyopathy		100%	3 of 3
MYH6		Atrial Septal Defect Ostium Secundum Type, Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	99.94%	140 of 142
MYH7	High Risk	Dilated Cardiomyopathy, Left Ventricular Noncompaction, Familial Hypertrophic Cardiomyopathy, Classic Multiminicore Myopathy, Ebstein Malformation, MYH7-Related Late-Onset Scapuloperoneal Muscular Dystrophy, Congenital Myopathy With Fiber-type Disproportion	AD,AR	99.95%	1053 of 1054
MYL2	High Risk	Cardiomyopathy, Familial Hypertrophic, Congenital Fiber-Type Disproportion Myopathy	AD	100%	67 of 67
MYL3	High Risk	Cardiomyopathy, Familial Hypertrophic	AD,AR	100%	42 of 42
MYL4		Familial Atrial Fibrillation	AD	100%	2 of 2
MYO18B		Autosomal Recessive Klippel-Feil Syndrome With Myopathy And Facial Dysmorphism	AR	99.39%	8 of 9
MYOT		Autosomal Dominant Limb-Girdle Muscular Dystrophy Type 1A, Distal Myotilinopathy Myopathy, Spheroid Body Myotilinopathy	AD	100%	17 of 17
MYPN		Cap Myopathy, Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Childhood-onset Nemaline Myopathy, Familial Isolated Restrictive Cardiomyopathy, Nemaline Myopathy	AD,AR	99.94%	49 of 49
MYRF		Cardiac-Urogenital Syndrome, Encephalitis/Encephalopathy With Reversible Myelin Vacuolization	AD	99.83%	27 of 27
ND2		Isolated Complex I Deficiency, Leber Hereditary Optic Neuropathy, Leber Optic Atrophy, Mitochondrial DNA-associated Leigh Syndrome	MI	85.56%	NA of NA
ND3		Isolated Complex I Deficiency, Mitochondrial DNA-associated Leigh Syndrome		99.99%	NA of NA
ND4		Leber Hereditary Optic Neuropathy, Leber Optic Atrophy, MELAS, Mitochondrial DNA-associated Leigh Syndrome	MI	na	na
ND4L		Leber Hereditary Optic Neuropathy, Leber Optic Atrophy	MI	99.83%	NA of NA
ND5		Leber Hereditary Optic Neuropathy, Leber Optic Atrophy, MELAS, MERRF, Mitochondrial DNA-associated Leigh Syndrome	MI	99.89%	NA of NA
ND6		Leber Hereditary Optic Neuropathy, Leber Optic Atrophy, MELAS, Mitochondrial DNA-associated Leigh Syndrome	MI	100%	NA of NA

NDUFAF2		Isolated Complex I Deficiency, Leigh Syndrome With Leukodystrophy, Mitochondrial Complex I Deficiency, Nuclear Type	AR	99.39%	6 of 6
NEXN		Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	99.70%	44 of 45
NF1		17q11.2 Microduplication Syndrome, Hereditary Pheochromocytoma-paraganglioma, Juvenile Myelomonocytic Leukemia, Neurofibromatosis Type 1 Due To Nf1 Mutation Or Intragenic Deletion, Neurofibromatosis-Noonan Syndrome, Familial Spinal Neurofibromatosis, Type I, Watson Syndrome	AD	97.97%	3082 of 3166
NKX2-5	High Risk	Atrial Septal Defect With Or Without Atrioventricular Conduction Defects, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Familial Bicuspid Aortic Valve, Familial Progressive Cardiac Conduction Defect, Hypoplastic Left Heart Syndrome, Congenital Nongoitrus Hypothyroidism, Tetralogy Of Fallot, Thyroid Ectopia, Ventricular Septal Defect	AD,AR	99.98%	112 of 116
NONO		Macrocephaly-Intellectual Disability, Left Ventricular Non Compaction Syndrome, X-linked Mental Retardation	X,XR,G	99.59%	NA of NA
NOS1AP		Romano-Ward Syndrome		100%	4 of 4
NRAP		Myofibrillar Myopathy, Reducing Body Myopathy 1A		99.98%	7 of 7
NRAS		Colorectal Cancer, Epidermal Nevus, Somatic Giant Pigmented Hairy Nevus, Large Congenital Melanocytic Nevus, Linear Nevus Sebaceus Syndrome, Neurocutaneous Melanosis, Noonan Syndrome, RAS-associated Autoimmune Lymphoproliferative Syndrome Type IV, Schimmelpenning-Feuerstein-Mims Syndrome, Nonmedullary Thyroid Cancer	AD	100%	15 of 15
NUP155		Familial Atrial Fibrillation	AR	99.91%	2 of 3
PARS2		Early Infantile Epileptic Encephalopathy	AR	100%	7 of 7
PCCA		Propionic Acidemia	AR	100%	137 of 137
PCCB		Propionic Acidemia	AR	99.95%	136 of 138
PKP2	High Risk	Familial Arrhythmogenic Right Ventricular Dysplasia, Brugada Syndrome	AD	100%	306 of 307
PLEC		Aplasia Cutis Congenita, Epidermolysis Bullos, Limb-Girdle Muscular Dystrophy Type 2Q	AD,AR	99.98%	113 of 113
PLEKHM2		Left Ventricular Non Compaction, Dilated Cardiomyopathy		99.94%	1 of 1
PLN	High Risk	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100%	26 of 33
PNPLA2		Neutral Lipid Storage Disease With Myopathy, Neutral Lipid Storage Myopathy	AR	100%	53 of 53
POMT1		Congenital Muscular Dystrophy With Cerebellar Involvement and Intellectual Dissability, Muscle-Eye-Brain Disease, Muscular Dystrophy-dystroglycanopathy Limb-Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	100%	105 of 105
PPA2		Sudden Cardiac Failure	AR	99.95%	9 of 9
PPCS		Dilated Cardiomyopathy	AR	98.95%	4 of 4
PPP1CB		Noonan Syndrome-like Disorder With Loose Anagen Hair	AD	99.87%	12 of 12
PRDM16		1p36 Deletion Syndrome, Familial Isolated Dilated Cardiomyopathy, Left Ventricular Noncompaction	AD	98.81%	20 of 20
PRKAG2	High Risk	Familial Hypertrophic Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff-Parkinson-White Syndrome	AD	99.98%	61 of 61
PTPN11	High Risk	Juvenile Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis, Noonan Syndrome	AD	100%	150 of 151
QRSL1		Combined Oxidative Phosphorylation Deficiency	AR	99.91%	6 of 7
RAF1		Dilatd Cardiomyopathy, Leopard Syndrome, Noonan Syndrome	AD	100%	64 of 64
RASA2		Noonan Syndrome		99.82%	5 of 5
RBCK1		Early Onset Polyglucosan Body Myopathy With Or Without Immunodeficiency	AR	100%	13 of 13
RBM20	High Risk	Dilated Cardiomyopathy	AD	96.83%	73 of 75
RIT1		Noonan Syndrome	AD	99.85%	27 of 27
RMND1		Combined Oxidative Phosphorylation Deficiency	AR	99.67%	15 of 16
RRAS		Noonan Syndrome		95.86%	3 of 3
RYR2	High Risk	Familial Arrhythmogenic Right Ventricular Dysplasia, Catecholaminergic Polymorphic Ventricular Tachycardia	AD	99.20%	466 of 472
SALL4		Acro-Renal-Ocular Syndrome, Duane Retraction Syndrome, Duane-Radial Ray Syndrome, Ivic Syndrome	AD	100%	54 of 54
SCN10A		Brugada Syndrome, Familial Episodic Pain Syndrome, Paroxysmal Extreme Pain Disorder, Primary Erythromelalgia, Romano-Ward Syndrome	AD	99.89%	96 of 96
SCN1B		Familial Atrial Fibrillation, Brugada Syndrome, Dravet Syndrome, Early Infantile Epileptic Encephalopathy, Familial Progressive Cardiac Conduction Defect, Generalized Epilepsy With Febrile Seizures	AD,AR	99.67%	46 of 48
SCN3B		Brugada Syndrome	AD	100%	7 of 7
SCN5A	High Risk	Familial Atrial Fibrillation, Brugada Syndrome, Dilated Cardiomyopathy, Familial Progressive Cardiac Conduction Defect, Long Qt Syndrome, Progressive Familial Heart Block, Romano-Ward Syndrome, Sick Sinus Syndrome, Sudden Infant Death Syndrome	AD,AR,MU	99.45%	929 of 942
SCNN1B		Generalized Bronchiectasis, Pseudohypoaldosteronism Type 1, Liddle Syndrome	AD,AR	100%	56 of 56
SCNN1G		Bronchiectasis With Or Without Elevated Sweat Chloride, Generalized Pseudohypoaldosteronism Type 1, Liddle	AD,AR	100%	28 of 28
SCO1		Mitochondrial Complex IV Deficiency	AR,MI	100%	6 of 6
SCO2		Autosomal Recessive Axonal Charcot-Marie-Tooth Disease Due To Copper Metabolism Defect, Cardioencephalomyopathy, Leigh Syndrome With Cardiomyopathy	AD,AR	100%	38 of 38
SDHA		Dilated Cardiomyopathy, Gastrointestinal Stromal Tumor, Hereditary Pheochromocytoma-pPraganglioma, Isolated Succinate-CoQ Reductase Deficiency, Leigh Syndrome, Mitochondrial Complex II Deficiency	AD,AR,MI	99.98%	103 of 103
SELENON		Classic Multiminicore Myopathy, Congenital Fiber-Type Disproportion Myopathy, Rigid Spine Muscular Dystrophy	AD,AR	89%	NA of NA

SGCA		Alpha-Sarcoglycan-Related Limb-girdle Muscular Dystrophy	AR	100%	119 of 119
SGCB		Beta-Sarcoglycan-Related Limb-girdle Muscular Dystrophy	AR	98.36%	55 of 65
SGCD		Dilated Cardiomyopathy, Delta-Sarcoglycan-Related Limb-Girdle Muscular Dystrophy	AD,AR	99.89%	31 of 32
SGCG		Gamma-Sarcoglycan-Related Limb-girdle Muscular Dystrophy	AR	100%	53 of 55
SHOC2		Noonan Syndrome-like Disorder With Loose Anagen Hair	AD	99.98%	8 of 8
SLC22A5		Systemic Primary Carnitine Deficiency	AR	100%	161 of 162
SLC25A20		Carnitine-Acylcarnitine Translocase Deficiency	AR	100%	39 of 39
SLC25A3		Cardiomyopathy-Hypotonia-lactic Acidosis Syndrome, Mitochondrial Phosphate Carrier Deficiency	AR	100%	6 of 6
SLC25A4		Autosomal Dominant Progressive External Ophthalmoplegia, Congenital Cataract-Hypertrophic Cardiomyopathy-mitochondrial Myopathy Syndrome, Mitochondrial DNA Depletion Syndrome 12 (Cardiomyopathic Type)	AD,AR	99.84%	16 of 16
SMCHD1		Bosma Arhinia Microphthalmia Syndrome, Facioscapulohumeral Muscular Dystrophy, Hyposmia-Nasal And Ocular Hypoplasia-hypogonadotropic Hypogonadism Syndrome	AD,MU,D	99.64%	131 of 137
SOS1		Hereditary Gingival Fibromatosis, Noonan Syndrome	AD	100%	103 of 104
SOS2		Noonan Syndrome	AD	99.48%	6 of 7
SPEG		Autosomal Recessive Centronuclear Myopathy	AR	99.26%	17 of 17
SPRED1		Legius Syndrome	AD	100%	84 of 84
STAG2		Alobar Holoprosencephaly, X-linked Neurodevelopmental Disorder With Craniofacial Abnormalities, Semilobar Holoprosencephaly, Microduplication Syndrome	X,XR,G	99.09%	NA of NA
TAB2		Multiple Types Congenital Heart Defects, Polyvalvular Heart Disease Syndrome	AD	99%	13 of 13
TAZ	High Risk	Barth Syndrome, Familial Isolated Dilated Cardiomyopathy	X,XR,G	100%	NA of NA
TBX20	High Risk	Atrial Septal Defect Ostium Secundum Type	AD	99.98%	33 of 34
TBX5		Holt-Oram Syndrome	AD	100%	143 of 152
TCAP		Familial Hypertrophic Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy, Limb-Girdle Muscular Dystrophy	AD,AR	100%	33 of 33
TECRL		Catecholaminergic Polymorphic Ventricular Tachycardia	AR	99.48%	4 of 4
TGFB3		Familial Arrhythmogenic Right Ventricular Dysplasia, Familial Thoracic Aortic Aneurysm And Aortic Dissection, Loeys-Dietz Syndrome 5	AD	100%	34 of 35
TMEM43	High Risk	Familial Arrhythmogenic Right Ventricular Dysplasia, Autosomal Dominant Emery-Dreifuss Muscular Dystrophy	AD	99.98%	26 of 26
TMEM70		Mitochondrial Complex V (ATP Synthase) Deficiency Nuclear Type, TMEM-70-related Mitochondrial Encephalo-cardio-myopathy	AR	100%	22 of 24
TNNC1	High Risk	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100%	28 of 28
TNNI3	High Risk	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Familial Restrictive Cardiomyopathy,	AD,AR	100%	139 of 139
TNNI3K		Cardiac Conduction Disease With Or Without Dilated Cardiomyopathy	AD	99.97%	4 of 4
TNNT2	High Risk	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Familial Restrictive Cardiomyopathy	AD	100%	169 of 169
TOR1AIP1		Limb-Girdle Muscular Dystrophy	AR	97.50%	5 of 6
TPM1	High Risk	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100%	108 of 108
TRDN		Catecholaminergic Polymorphic Ventricular Tachycardia, Romano-Ward Syndrome	AD,AR	98.72%	10 of 12
TRIM32		Bardet-Biedl Syndrome, Limb-girdle Muscular Dystrophy	AR	100%	17 of 17
TRNC		Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-likeepisodes (MELAS)	MI	na	na
TRNE		Maternally-Inherited Diabetes And Deafness, Mitochondrial Myopathy With Reversible Cytochrome C Oxidase Deficiency		na	na
TRNF		MELAS, Myoclonic Epilepsy Associated With Ragged-red Fibers (MERRF)	MI	na	na
TRNH		MELAS, MERRF		na	na
TRNI		Myoclonic Epilepsy Associated With Ragged-red Fibers	MI	na	na
TRNK		Maternally-Inherited Diabetes And Deafness, MERRF, Mitochondrial DNA-associated Leigh Syndrome, Mitochondrial Dna-related Cardiomyopathy And Hearing Loss, MELAS, MERRF	MI	na	na
TRNL1		Kearns-Sayre Syndrome, Maternally-Inherited Diabetes And Deafness, MELAS, MERRF, Mitochondrial DNA-associated Leigh Syndrome, Mitochondrial DNA-related Progressive External Ophthalmoplegia	MI	na	na
TRNL2		Mitochondrial DNA-related Progressive External Ophthalmoplegia		na	na
TRNN		Mitochondrial Complex IV Deficiency, Mitochondrial DNA-Related Progressive External Ophthalmoplegia	AR,MI	na	na
TRNQ		MELAS, MERRF	MI	na	na
TRNS1		Aminoglycoside-Induced Deafness, MELAS, MERRF, Mitochondrial Complex IV Deficiency, Mitochondrial DNA-related Progressive External Ophthalmoplegia, Palmoplantar Keratoderma-Deafness Syndrome	AR,MI	na	na
TRNS2		MELAS, MERRF	MI	na	na
TRNT		Lethal Infantile Mitochondrial Myopathy	MI	na	na
TRNV		Mitochondrial DNA-associated Leigh Syndrome, MELAS	MI	na	na
TRNW		MELAS, Mitochondrial DNA-associated Leigh Syndrome	AR,MI	na	na
TRPM4		Brugada Syndrome, Erythrokeratodermia Veriabilis Et Progressiva, Familial Progressive Cardiac Conduction Defect, Progressive Familial Heart Block	AD	99.98%	44 of 44
TSFM		Combined Oxidative Phosphorylation Deficiency	AR	93.35%	11 of 14
TTN	High Risk	Autosomal Recessive Centronuclear Myopathy, Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Classic Multiminicore Myopathy, Limb-Girdle Muscular Dystrophy, Early-Onset Myopathy With Fatal Cardiomyopathy, Myofibrillar Myopathy, Tibial Muscular Dystrophy	AD,AR	97.93%	1153 of 1219

TTR	High Risk	Amyloidosis VII, Carpal Tunnel Syndrome	AD	100%	195 of 196
VARS2		Combined Oxidative Phosphorylation Deficiency	AR	99.90%	16 of 16
VCL		Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	99.99%	36 of 37
VCP		Adult-Onset Distal Myopathy Due To VCP Mutation, Amyotrophic Lateral Sclerosis, Autosomal Dominant Charcot-Marie-Tooth Disease Type 2Y, Early-Onset Paget Disease And Frontotemporal Dementia, Progressive Non-fluent Aphasia, Spastic Paraplegia-paget Disease Of Bone Syndrome	AD	100%	68 of 69
VPS13A		Choreoacanthocytosis	AR	99.37%	120 of 122
XK		Mcleod Syndrome	X,G	99.97%	NA of NA

* Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial
** HGMD: Number of clinically relevant mutations according to HGMD

Methodology

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References

1. 2019 ACC/AHA Guideline on the Primary Prevention of Cardiovascular Disease: A Report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. J Am Coll Cardiol 2019;March 17
2. Schwartz, P. J., Ackerman, M. J., George, A. L., Jr, & Wilde, A. (2013). Impact of genetics on the clinical management of channelopathies. *Journal of the American College of Cardiology*, *62*(3), 169–180. https://doi.org/10.1016/j.jacc.2013.04.044
3. Lüscher T. F. (2019). Channelopathies and sudden cardiac death: genetics and pharmacological triggers. *European heart journal*, *40*(37), 3067–3070. https://doi.org/10.1093/eurheartj/ehz710
4. Burke, M. A., Cook, S. A., Seidman, J. G., & Seidman, C. E. (2016). Clinical and Mechanistic Insights Into the Genetics of Cardiomyopathy. *Journal of the American College of Cardiology*, *68*(25), 2871–2886. https://doi.org/10.1016/j.jacc.2016.08.079
5. Hershberger, R. E., Givertz, M. M., Ho, C. Y., Judge, D. P., Kantor, P. F., McBride, K. L., Morales, A., Taylor, M., Vatta, M., Ware, S. M., & ACMG Professional Practice and Guidelines Committee (2018). Genetic evaluation of cardiomyopathy: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genetics in medicine : official journal of the American College of Medical Genetics*, *20*(9), 899–909. https://doi.org/10.1038/s41436-018-0039-z
6. Hershberger, R. E., Givertz, M. M., Ho, C. Y., Judge, D. P., Kantor, P. F., McBride, K. L., Morales, A., Taylor, M., Vatta, M., & Ware, S. M. (2018). Genetic Evaluation of Cardiomyopathy-A Heart Failure Society of America Practice Guideline. *Journal of cardiac failure*, *24*(5), 281–302. https://doi.org/10.1016/j.cardfail.2018.03.004
7. Corrado, D., Basso, C., & Judge, D. P. (2017). Arrhythmogenic Cardiomyopathy. *Circulation research*, *121*(7), 784–802. https://doi.org/10.1161/CIRCRESAHA.117.309345