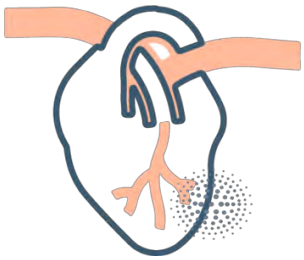
Cardiomyopathy Precision Panel



Overview

Cardiomyopathies are 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).

The Igenomix Cardiomyopathy Precision Panel serves as a diagnostic and tool ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes.

Indications

The Igenomix Cardiomyopathy Precision Panel is indicated in those cases where there is a clinical suspicion of cardiomyopathy with or without the following manifestations:

- 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.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AARS2	Combined Oxidative Phosphorylation Deficiency	AR	100	54 OF 54
ABCC9	Familial Atrial Fibrillation, Dilated Cardiomyopathy, Brugada Syndrome	AD	100	51 OF 51
ACAD9	Acyl-CoAa Dehydrogenase Deficiency	AR	100	62 OF 62
ACADVL	Acyl-CoA Dehydrogenase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency	AR	100	329 OF 329
ACTA1	Congenital Myopathy With Fiber-Type Disproportion , Nemaline Myopathy	AD,AR	100	224 OF 224
ACTC1	Atrial Septal Defect, Dilated Cardiomyopathy, Left Ventricular Noncompaction, Familial Hypertrophic Cardiomyopathy	AD	99.93	72 OF 74
ACTN2	Dilated Cardiomyopathy With Or Without Left Ventricular Noncompaction, Congenital Myopathy With Structured Cores And Z-line Abnormalities	AD	100	56 OF 56
AGL	Glycogen Storage Disease III	AR	100	253 OF 253
ALMS1	Alstrom Syndrome	AR	99.92	302 OF 305
ALPK3	Familial Hypertrophic Cardiomyopathy	AR	97.29	7 OF 7
ANO5	Miyoshi Muscular Dystrophy, Limb-Girdle Muscular Dystrophy	AD,AR	99.78	171 OF 173
APOA1	Familial Visceral Amyloidosis, Apolipoprotein A-1 Deficiency	AD	99.89	68 OF 70
ATP6	Leber Optic Atrophy , Neuropathy, Ataxia, And Retinitis Pigmentosa, Mitochondrial DNA-Associated Leigh Syndrome, Narp Syndrome	MI	NA	NA
ATP8	Kearns-Sayre Syndrome		98.02	NA OF NA
BAG3	Dilated Cardiomyopathy	AD	100	83 OF 85
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Noonan Syndrome	AD	100	80 OF 80
CACNA1C	Brugada Syndrome, Timothy Syndrome, Romano-Ward Syndrome	AD	99.8	85 OF 85
CALR3	Hypertrophic Cardiomyopathy, Restrictive Cardiomyopathy		100	5 OF 5
CAPN3	Limb-Girdle Muscular Dystrophy	AD,AR	100	503 OF 505
CASQ2	Catecholaminergic Polymorphic Ventricular Tachycardia With Orwithout Atrial Dysfunction And/Or Dilated Cardiomyopathy	AD,AR	100	39 OF 40
CBL	Noonan Syndrome	AD	100	46 OF 47
CDH2	Familial Arrhythmogenic Right Ventricular Dysplasia	AD	99.98	16 OF 16
CHRM2	Dilated Cardiomyopathy		99.98	1 OF 1
COX15	Fatal Infantile Cardioencephalomyopathy, Leigh Syndrome	AR,MI	100	5 OF 5
COX3	Leber Optic Atrophy , Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes , Leber Hereditary Optic Neuropathy	MI	NA	NA
CPT2	Carnitine Palmitoyltransferase II Deficiency	AD,AR	99.99	116 OF 116
CRPPA	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Limb-Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	97.69	NA OF NA
CRYAB	Dilated Cardiomyopathy	AD,AR	100	30 OF 30
CSRP3	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100	36 OF 36

CTNNA3	Familial Arrhythmogenic Right Ventricular Dysplasia	AD	99.97	14 OF 17
CYTB	Histiocytoid Cardiomyopathy	MI	98.8	NA OF NA
DBH	Dopamine Beta-Hydroxylase Deficiency,	AR	100	11 OF 11
DES	Dilated Cardiomyopathy	AD,AR	99.97	133 OF 134
DMD	Dilated Cardiomyopathy	X,XR,G	99.96	NA OF NA
DNAJC19	Dilated Cardiomyopathy With Ataxia	AR	100	6 OF 6
DOLK	Familial Isolated Dilated Cardiomyopathy	AR	99.98	13 OF 13
DPM3	Congenital Disorder Of Glycosylation Type Io, Muscular Dystrophy-Dystroglycanopathy (Congenital With Impaired Intellectual Development)	AR	100	4 OF 4
DSC2	Familial Arrhythmogenic Right Ventricular Dysplasia	AD,AR	100	123 OF 124
DSG2	Familial Arrhythmogenic Right Ventricular Dysplasia, Dilated Cardiomyopathy	AD	99.38	167 OF 169
DSP	Familial Arrhythmogenic Right Ventricular Dysplasia, Dilated Cardiomyopathy	AD,AR	99.91	366 OF 369
DTNA	Left Ventricular Noncompaction	AD	97	10 OF 10
DYSF	Miyoshi Myopathy, Limb-Girdle Muscular Dystrophy	AR	100	604 OF 606
EEF1A2	Early Infantile Epileptic Encephalopathy	AD	100	14 OF 14
ELAC2	Combined Oxidative Phosphorylation Deficiency	AR	100	32 OF 32
EMD	X-linked Emery-Dreifuss Muscular Dystrophy	X,XR,G	99.92	NA OF NA
EPG5	Immunodeficiency With Cleft Lip/Palate, Cataract, Hypopigmentation and Absent Corpus Callosum , Vici 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
EYA4	Dilated Cardiomyopathy	AD	100	32 OF 32
FBXL4	Mitochondrial DNA Depletion Syndrome (Encephalomyopathic Type)	AR	99.26	46 OF 51
FBXO32	Dilated Cardiomyopathy		100	2 OF 2
FHL1	Reducing Body Myopathy, X-Linked Emery-Dreifuss Muscular Dystrophy	X,XR,XD,G	99.98	NA OF NA
FHOD3	Hypertrophic Cardiomyopathy		99.95	35 OF 35
FKRP	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Limb-Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	99.9	157 OF 157
FKTN	Dilated Cardiomyopathy, Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Limb-Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	98	54 OF 56
FLNC	Familial Hypertrophic Cardiomyopathy, Familial Isolated Restrictive Cardiomyopathy	AD	100	185 OF 186
FOXRED1	Mitochondrial Complex I Deficiency, Isolated Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	100	13 OF 13
GAA	Glycogen Storage Disease II	AR	100	623 OF 624
GATA6	Atrial Septal Defect, Atrioventricular Septal Defect, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Tetralogy Of Fallot	AD,AR	84.19	66 OF 84
GATAD1	Dilated Cardiomyopathy	AR	88.2	1 OF 1
GBE1	Glycogen Storage Disease IV, Adult Polyglucosan Body Disease	AR	99.95	71 OF 74
GFM1	Combined Oxidative Phosphorylation Deficiency	AR	100	27 OF 27
GLA	Fabry Disease	X,XR,G	98	NA OF NA
GLB1	GM1-Gangliosidosis, Morquio Syndrome	AR	100	242 OF 243
GMPPB	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Congenital Myasthenic Syndromes With Glycosylation Defect, Limb-Girdle Muscular Dystrophy	AR	99.95	53 OF 53
GSK3B	Usher Syndrome, Alzheimer Disease		99.91	1 OF 1
GTPBP3	Combined Oxidative Phosphorylation Deficiency	AR	99.94	17 OF 17
HADHA	Long-Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency, Mitochondrial Trifunctional Protein Deficiency	AR	100	75 OF 75
HAND1	Hypoplastic Left Heart Syndrome		99.89	9 OF 9
HCN4	Brugada Syndrome	AD	98.01	40 OF 41
HFE	Alzheimer Disease, Hemochromatosis	AD,AR	100	55 OF 57
HRAS	Costello Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome	AD	100	34 OF 34
IDUA	Hurler Syndrome, Hurler-Scheie Syndrome	AR	99.73	287 OF 292
ILK	Dilated Cardiomyopathy		100	14 OF 14

JPH2	Familial Hypertrophic Cardiomyopathy	AD	98.24	17 OF 17
JUP	Familial Arrhythmogenic Right Ventricular Dysplasia	AD,AR	100	56 OF 56
KLHL24	Epidermolysis Bullosa Simplex, Generalized, With Scarring And Hair Loss	AD	99.96	8 OF 8
KRAS	Cardiofaciocutaneous Syndrome, Noonan Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 OF 38
LAMA2	Congenital Merosin-Deficient Muscular Dystrophy, Limb-Girdle Muscular Dystrophy	AR	100	363 OF 377
LAMP2	Danon Disease , Glycogen Storage Disease Due To Lamp-2 Deficiency	X,XD,G	99.96	NA OF NA
LARGE1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Walker-Warburg Syndrome	AR	100	NA OF NA
LDB3	Dilated Cardiomyopathy, Dilated With Or Without Left Ventricular Noncompaction	AD	100	60 OF 60
LEMD2	Congenital Cataract, Juvenile	AR	93.48	3 OF 3
LMNA	Dilated Cardiomyopathy, Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Dilated Cardiomyopathy-Hypergonadotropic Hypogonadism	AD,AR	100	619 OF 620
LMOD2	Familial Hypertrophic Cardiomyopathy		99.37	1 OF 1
LRRC10	Dilated Cardiomyopathy		100	5 OF 5
LZTR1	Noonan Syndrome	AD	99.99	136 OF 136
MAP2K1	Cardiofaciocutaneous Syndrome, Noonan Syndrome	AD	100	31 OF 31
MAP2K2	Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome	AD	100	37 OF 37
MAP3K8	Lung Cancer, Rheumatoid Arthritis	AD	99.91	1 OF 1
MLYCD	Malonyl-CoA Decarboxylase Deficiency	AR	93.84	32 OF 40
MRAS	Noonan Syndrome	AD	100	3 OF 3
MT-CO1	Mitochondrial Complex IV Deficiency		97.64	NA OF NA
MT-CO2	Mitochondrial Complex IV Deficiency		99.19	NA OF NA
MT-ND1	Mitochondrial Myopathy		98.8	NA OF NA
MTO1	Combined Oxidative Phosphorylation Deficiency	AR	99.83	31 OF 31
MYBPC3	Familial Hypertrophic Cardiomyopathy, Left Ventricular Noncompaction, Dilated Cardiomyopathy	AD,AR	99.95	1072 OF 1079
MYBPHL	Dilated Cardiomyopathy		100	3 OF 3
MYH6	Atrial Septal Defect, Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	99.94	140 OF 142
MYH7	Dilated Cardiomyopathy, Left Ventricular Noncompaction, Familial Hypertrophic Cardiomyopathy	AD,AR	99.95	1053 OF 1054
MYL2	Familial Hypertrophic Cardiomyopathy, Congenital Fiber-type Disproportion Myopathy	AD	100	67 OF 67
MYL3	Familial Hypertrophic Cardiomyopathy	AD,AR	100	42 OF 42
MYL4	Familial Atrial Fibrillation	AD	100	2 OF 2
MYLK3	Hypoplastic Right Heart Syndrome, Dilated Cardiomyopathy		99.83	3 OF 3
MYOT	Myopathy Spheroid Body Myotilinopathy, Limb-Girdle Muscular Dystrophy	AD	100	17 OF 17
MYPN	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Restrictive Cardiomyopathy	AD,AR	99.94	49 OF 49
MYRF	Cardiac-Urogenital Syndrome	AD	99.83	27 OF 27
ND2	Leber Optic Atrophy , Isolated Complex I Deficiency , Leber Hereditary Optic Neuropathy , 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 Optic Atrophy, Leber Hereditary Optic Neuropathy, Mitochondrial DNA-Associated Leigh Syndrome	MI	NA	NA
ND4L	Leber Optic Atrophy, Leber Hereditary Optic Neuropathy	MI	99.83	NA OF NA
ND5	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Leber Hereditary Optic Neuropathy, Mitochondrial DNA-Associated Leigh Syndrome	MI	99.89	NA OF NA
ND6	Leber Optic Atrophy , Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Leber Hereditary Optic Neuropathy, Mitochondrial DNA-Associated Leigh Syndrome	MI	100	NA OF NA
NDUFAF2	Mitochondrial Complex I Deficiency, Isolated Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	99.39	6 OF 6
NEXN	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	99.7	44 OF 45
NF1	Neurofibromatosis-Noonan Syndrome, Watson Syndrome	AD	97.97	3082 OF 3166
NKX2-5	Atrial Septal Defect With Or Without Atrioventricular Conduction Defects, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Hypoplastic Left Heart Syndrome, Tetralogy Of Fallot, Ventricular Septal Defect	AD,AR	99.98	112 OF 116

NONO	Macrocephaly-Intellectual Disability-Left Ventricular Non Compaction Syndrome	X,XR,G	99.59	NA OF NA
NRAP	Myofibrillar Myopathy, Reducing Body Myopathy, Congenital Myasthenic Syndrome		99.98	7 OF 7
NRAS	Noonan Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome	AD	100	15 OF 15
PCCA	Propionic Acidemia	AR	100	137 OF 137
PCCB	Propionic Acidemia	AR	99.95	136 OF 138
PKP2	Familial Arrhythmogenic Right Ventricular Dysplasia, Brugada Syndrome	AD	100	306 OF 307
PLEC	Epidermolysis Bullosa Simplex With Muscular Dystrophy, Limb-Girdle Muscular Dystrophy	AD,AR	99.98	113 OF 113
PLEKHM2	Leukodystrophy and Acquired Microcephaly With or Without Dystonia		99.94	1 OF 1
PLN	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100	26 OF 33
PNPLA2	Neutral Lipid Storage Disease With Myopathy	AR	100	53 OF 53
PPA2	Sudden Cardiac Failure, 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	Left Ventricular Noncompaction, Dilated Cardiomyopathy	AD	98.81	20 OF 20
PRKAG2	Familial Hypertrophic Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff-Parkinson-White Syndrome ,	AD	99.98	61 OF 61
PTPN11	Leopard Syndrome, Noonan Syndrome	AD	100	150 OF 151
QRSL1	Combined Oxidative Phosphorylation Deficiency	AR	99.91	6 OF 7
RAF1	Dilated Cardiomyopathy, Leopard Syndrome, Noonan Syndrome	AD	100	64 OF 64
RASA1	Capillary Malformation-Arteriovenous Malformation, Parkes Weber Syndrome	AD	99.56	169 OF 169
RASA2	Noonan Syndrome		99.82	5 OF 5
RBCK1	Polyglucosan Body Myopathy, Early-Onset, With Or Without Immunodeficiency	AR	100	13 OF 13
RBM20	Dilated Cardiomyopathy	AD	96.83	73 OF 75
RIT1	Noonan Syndrome 8	AD	99.85	27 OF 27
RRAS	Noonan Syndrome		95.86	3 OF 3
RYR2	Familial Arrhythmogenic Right Ventricular Dysplasia, Catecholaminergic Polymorphic Ventricular Tachycardia With Or Without Atrial Dysfunction And/Or Dilated Cardiomyopathy	AD	99.2	466 OF 472
SCN5A	Familal Atrial Fibrillation, Brugada Syndrome, Dilated Cardiomyopathy, Long QT Syndrome, Progressive Familial Heart Block, Type Ia, Sick Sinus Syndrome, Sudden Infant Death Syndrome , Ventricular Fibrillation During Myocardial Infarction, Romano-Ward Syndrome	AD,AR,MU	99.45	929 OF 942
SCNN1B	Bronchiectasis, Liddle Syndrome, Pseudohypoaldosteronism Type I	AD,AR	100	56 OF 56
SCNN1G	Bronchiectasis With Or Without Elevated Sweat Chloride, Liddle Syndrome, Pseudohypoaldosteronism Type I	AD,AR	100	28 OF 28
SCO1	Mitochondrial Complex IV Deficiency	AR,MI	100	6 OF 6
SCO2	Fatal Infantile Cardioencephalomyopathy, Autosomal Recessive Axonal Charcot-Marie-Tooth Disease Due To Copper Metabolism Defect, Leigh Syndrome With Cardiomyopathy	AD,AR	100	38 OF 38
SDHA	Dilated Cardiomyopathy, Leigh Syndrome, Mitochondrial Complex II Deficiency, Isolated Succinate-CoQ Reductase Deficiency, Leigh Syndrome With Leukodystrophy	AD,AR,MI	99.98	103 OF 103
SGCA	Limb-Girdle Muscular Dystrophy, Alpha-Sarcoglycan-Related Limb-Girdle Muscular Dystrophy	AR	100	119 OF 119
SGCB	Limb-Girdle Muscular Dystrophy, Beta-Sarcoglycan-Related Limb-Girdle Muscular Dystrophy	AR	98.36	55 OF 65
SGCD	Dilated Cardiomyopathy, Limb-Girdle Muscular Dystrophy, Delta-Sarcoglycan-Related, Familial Isolated Dilated Cardiomyopathy	AD,AR	99.89	31 OF 32
SGCG	Limb-Girdle Muscular Dystrophy, 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
SLC25A4	Mitochondrial DNA Depletion Syndrome 12 (Cardiomyopathic Type) Congenital Cataract-Hypertrophic Cardiomyopathy-Mitochondrial Myopathy Syndrome	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	Noonan Syndrome, Hereditary Gingival Fibromatosis	AD	100	103 OF 104
SOS2	Noonan Syndrome	AD	99.48	6 OF 7
SPEG	Centronuclear Myopathy	AR	99.26	17 OF 17
SPRED1	Legius Syndrome	AD	100	84 OF 84
TAB2	Congenital Heart Defects, Polyvalvular Heart Disease Syndrome	AD	99	13 OF 13
TAZ	Barth Syndrome, Familial Isolated Dilated Cardiomyopathy	X,XR,G	100	NA OF NA
TBX20	Atrial Septal Defect	AD	99.98	33 OF 34
TBX5	Holt-Oram Syndrome	AD	100	143 OF 152
TCAP	Familial Hypertrophic Cardiomyopathy, Limb-Girdle Muscular Dystrophy, Familial Isolated Dilated Cardiomyopathy	AD,AR	100	33 OF 33
TGFB3	Familial Arrhythmogenic Right Ventricular Dysplasia, Loeys-Dietz Syndrome, Familial Thoracic Aortic Aneurysm And Aortic Dissection	AD	100	34 OF 35
TMEM43	Familial Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular Dystrophy, Autosomal Dominant Emery-Dreifuss Muscular Dystrophy	AD	99.98	26 OF 26
TMEM70	Mitochondrial Complex V (ATP Synthase) Deficiency, TMEM-70 Mitochondrial Encephalo-Cardio-Myopathy	AR	100	22 OF 24
TNNC1	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100	28 OF 28
TNNI3	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	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Familial Restrictive Cardiomyopathy	AD	100	169 OF 169
TOR1AIP1	Limb-Girdle Muscular Dystrophy	AR	97.5	5 OF 6
TPM1	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100	108 OF 108
TRIM32	Bardet-Biedl Syndrome, Limb-Girdle Muscular Dystrophy	AR	100	17 OF 17
TRNC	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes	MI	NA	NA
TRNE	Mitochondrial Myopathy With Reversible Cytochrome C Oxidase Deficiency		NA	NA
TRNF	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Myoclonic Epilepsy Associated With Ragged-Red Fibers	MI	NA	NA
TRNH	MELAS, MERRF		NA	NA
TRNI	Myoclonic Epilepsy Associated With Ragged-Red Fibers	MI	NA	NA
TRNK	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Myoclonic Epilepsy Associated With Ragged-Red Fibers, Mitochondrial DNA-associated Leigh Syndrome	MI	NA	NA
TRNL1	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Myoclonic Epilepsy Associated With Ragged-Red Fibers, Kearns-Sayre Syndrome, Mitochondrial DNA-Associated Leigh Syndrome	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	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes	MI	NA	NA
TRNS1	Mitochondrial Complex IV Deficiency , Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Palmoplantar Keratoderma-Deafness Syndrome	AR,MI	NA	NA
TRNS2	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Usher Syndrome	MI	NA	NA
TRNT	Lethal Infantile Mitochondrial Myopathy	MI	NA	NA
TRNV	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial DNA-Associated Leigh Syndrome	MI	NA	NA
TRNW	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial Myopathy, Episodic, With Optic Atrophy And Reversible Leukoencephalopathy, Mitochondrial DNA-Associated Leigh Syndrome	AR,MI	NA	NA
TTN	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Limb Girdle Muscular Dystrophy, Early-Onset Myopathy With Fatal Cardiomyopathy	AD,AR	97.93	1153 OF 1219
TTR	Amyloidosis VII, Carpal Tunnel Syndrome	AD	100	195 OF 196

VCL	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	99.99	36 OF 37
VCP	Amyotrophic Lateral Sclerosis With Or Without Frontotemporal Dementia , Charcot-Marie-Tooth Disease, Inclusion Body Myopathy With Paget Disease Of Bone And Frontotemporal Dementia	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

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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