Congenital Muscular Dystrophies and Myopathies

Precision Panel

Overview

Congenital Muscular Dystrophies are an inherited group of progressive myopathic disorders resulting from defects in a number of genes responsible for normal muscle function, resulting in progressive muscle weakness without a central or peripheral nerve abnormality. The genes responsible for these diseases are specific muscle proteins that allow for proper contraction and relaxation of the muscles. Muscular dystrophies are classified according to the clinical phenotype, pathology and mode of inheritance. Inheritance pattern includes X-lined, autosomal recessive and autosomal dominant. Some examples include:

- X-linked: Duchenne, Becker, Emery-Dreifuss
- Autosomal dominant: Facioscapulohumeral, distal, ocular, oculopharyngeal
- Autosomgal recessive: Limb-girdle form

Congenital Myopathies are a group of genetic diseases that predominantly affect the muscles. The typical features can be found in neonates and infants, children or even adults. The classification of congenital myopathies follows a genetic criterion. However, the genotype-phenotype correlation remains variable and overlapping with congenital muscular dystrophies. Some examples of congenital myopathies include Nemaline Myopathy, Central Core Disease and Multiminicore Disease amongst others. Both congenital myopathies and muscular dystrophies carry a high risk of developing restrictive lung disease and orthopedic deformities.

The Igenomix Congenital Muscular Dystrophies and Myopathies Precision Panel can be used as a tool for an accurate diagnosis and differential diagnosis of muscle weakness 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 involved, and their high or intermediate penetrance.

Indications

The Igenomix Congenital Muscular Dystrophies and Myopathies Precision Panel is used for patients with a clinical suspicion or diagnosis with or without the following symptoms:

- Early-onset muscle weakness
- Decreased muscle tone
- Hypoactive deep tendon reflexes
- Delayed motor milestones
- Muscle atrophy
- Abnormally fixed joints
- Muscle deformities and contractures

- Family history of congenital myopathy or muscle dystrophy

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment involving a multidisciplinary team focusing on intensive physiotherapy and rehabilitation, bracing and surgical interventions and medical care to prevent complications and improve symptoms.
- Risk assessment of asymptomatic family members according to the mode of inheritance via genetic counselling.
- Improvement of delineation of genotype-phenotype correlation given the variability of severity and course of disease.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABHD5	Chanarin-Dorfman Syndrome, Neutral Lipid Storage Disease, Ichthyosis	AR	99.98	37 of 37
ACAD9	Acyl-Coa Dehydrogenase Deficiency	AR	100	62 of 62
ACADVL	Acyl-Coa Dehydrogenase Deficiency	AR	100	329 of 329
ACTA1	Multiple Myopathy Types, Rigid Spine Syndrome	AD,AR	100	224 of 224
ACTN2	Cardiomyopathy, Left Ventricular Noncompaction, Myopathy	AD	100	56 of 56
ADSS1	Myopathy	AR	86.52	3 of 3
AGL	Glycogen Storage Disease, Glycogen Debranching Enzyme Deficiency	AR	100	253 of 253
ANO5	Gnathodiaphyseal Dysplasia, Miyoshi Muscular Dystrophy, Limb-Girdle Muscular Dystrophy, Distal Anoctaminopathy	AD,AR	99.78	171 of 173
ATP2A1	Brody Myopathy	AR	100	20 of 20
B3GALNT2	Muscular Dystrophy-Dystroglycanopathy, Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	97.14	17 of 17
B4GAT1	Muscular Dystrophy-Dystroglycanopathy, Walker-Warburg Syndrome	AR	-	-
BAG3	Cardiomyopathy, Myopathy	AD	100	83 of 85
BICD2	Spinal Muscular Atrophy	AD	99.94	39 of 39
BIN1	Myopathy	AR	100	20 of 20
BVES	Limb Girdle Muscular Dystrophy	AR	99.47	2 of 2
CACNA1H	Hyperaldosteronism, Epilepsy	AD	98.05	71 of 71
CACNA1S	Hypokalemic And Thyrotoxic Periodic Paralysis, Hyperthermia	AD	100	64 of 64
CAPN3	Limb Girdle Muscular Dystrophy	AD,AR	100	503 of 505
CASQ1	Myopathy, Tubular Aggregate And Vacuolar Myopathy,	AD	100	6 of 6
CAV3	Cardiomyopathy, Creatine Phosphokinase, Long Qt Syndrome, Myopathy, Rippling Muscle Disease, Romano-Ward Syndrome	AD	100	50 of 50
CAVIN1	Berardinelli-Seip Lipodystrophy	AR	99.82	-
CCDC78	Myopathy	AD	100	5 of 5
CFL2	Nemaline Myopathy	AR	99.98	9 of 9
CHKB	Muscular Dystrophy	AR,MI	100	29 of 29

CLCN1	Myotonia Congenita, Thomsen And Becker Disease	AD,AR	100	321 of 321
CNTN1	Myopathy	AR	100	1 of 1
COL12A1	Bethlem Myopathy, Ullrich Muscular Dystrophy, Ehlers-Danlos Syndrome	AD	99.97	18 of 19
COL4A1	Angiopathy, Nephropathy, Aneurysms, Muscle Cramps, Microangiopathy, Leukoencephalopathy, Porencephaly, Hanac Syndrome, Walker-Warburg Syndrome	AD	99.99	173 of 173
COL4A2	Porencephaly	AD	99.93	28 of 28
COL6A1	Bethlem Myopathy, Ullrich Muscular Dystrophy	AD,AR	99.96	182 of 186
COL6A2	Bethlem Myopathy, Myosclerosis, Ullrich Muscular Dystrophy	AD,AR	100	223 of 225
COL6A3	Bethlem Myopathy, Dystonia, Ullrich Muscular Dystrophy	AD,AR	99.63	232 of 232
COX6A2	Mitochondrial Complex IV Deficiendy, Cardioencephalomyopathy	-	100	2 of 2
CPT2	Carnitine Palmitoyl Transferase Ii Deficiency, Encephalopathy	AD,AR	99.99	116 of 116
CRPPA	Muscular Dystrophy-Dystroglycanopathy, Walker-Warburg Syndrome	AR	97.69	-
CRYAB	Alpha-B Crystallinopathy, Cardiomyopathy, Cataract, Myopathy	AD,AR	100	30 of 30
DAG1	Muscular Dystrophy-Dystroglycanopathy, Muscle-Eye-Brain Disease, Multicystic Leucodystrophy, Walker-Warburg Syndrome	AR	99.98	9 of 9
DES	Cardiomyopathy, Myopathy, Scapuloperoneal Syndrome, Desminopathy	AD,AR	99.97	133 of 134
DMD	Cardiomyopathy, Becker Muscular Dystrophy, Duchenne Muscular Dystrophy, Intellectual Disability	X,XR,G	99.96	-
DNA2	Ophthalmoplegia, Seckel Syndrome, Mitochondrial Dna Deletion Syndrome	AD,AR	99.74	16 of 16
DNAJB6	Limb Girdle Muscular Dystrophy	AD	100	30 of 30
DNM2	Charcot-Marie-Tooth Disease, Contracture Syndrome, Myopathy	AD,AR	99	57 of 57
DPM1	Congenital Disorder Of Glycosylation	AR	97.25	9 of 9
DPM2	Congenital Disorder Of Glycosylation, Congenital Muscular Dystrophy, Intellectual Disability, Epilepsy	AR	99.87	2 of 2
DPM3	Congenital Disorder Of Glycosylation, Muscular Dystrophy-Dystroglycanopathy	AR	100	4 of 4
DUX4	Facioscapulohumeral Dystrophy	-	0	-
DYSF	Miyoshi Myopathy, Limb Girdle Muscular Dystrophy	AR	100	604 of 606
EMD	Emery-Dreifuss Muscular Dystrophy	X,XR,G	99.92	-
ENO3	Glycogen Storage Disease	AR	100	7 of 7
ETFA	Acyl-Coa Dehydrogenase Deficiency	AR	92.33	32 of 32
ETFB	Acyl-Coa Dehydrogenase Deficiency	AR	100	21 of 21
ETFDH	Acyl-Coa Dehydrogenase Deficiency	AR	100	221 of 222
FDX2	Mitochondrial Myopathy, Optic Atrophy, Leukoencephalopathy	AR,MI	100	-
FHL1	Myopathy, Uruguay Faciocardiomusculoskeletal Syndrome, Muscular Dystrophy	X,XR,XD,G	99.98	-
FKBP14	Ehlers-Danlos Syndrome, Progressive Kyphoscoliosis, Myopathy, Hearing Loss	AR	99.98	7 of 8
FKRP	Muscular Dystrophy-Dystroglycanopathy, Limb-Girdle Muscular Dystrophy, Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.9	157 of 157
FKTN	Cardiomyopathy, Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	98	54 of 56
FLAD1	Lipid Storage Myopathy	AR	97.13	13 of 14
FLNC	Cardiomyopathy, Filaminopathy, Myopathy	AD	100	185 of 186
FXR1	Myopathy, Respiratory Insufficiency, Bone Fractures	AR	99.93	1 of 1
GAA	Glycogen Storage Disease	AR	100	623 of 624
GBE1	Glycogen Storage Disease, Polyglucosan Body Disease	AR	99.95	71 of 74

GFER	Myopathy, Cataract, Hearing Loss, Developmental Delay	AR	99.89	6 of 6
GMPPB	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Intellectual Disability, Myasthenic Syndromes, Glycosylation Defect, Muscle-Eye-Brain Disease	AR	99.95	53 of 53
GNE	Nonaka Myopathy, Sialuria, Gne Myopathy, Sialuria	AD,AR	99.97	248 of 253
GOLGA2	Smith-Mccort Dysplasia, Mucolipidosis, Vohwinkel Syndrome, Dyggve-Melchior-Clausen Disease, Encephalopathy	-	99.89	3 of 3
GOSR2	Epilepsy	AR	88.39	6 of 6
GYG1	Glycogen Storage Disease, Polyglucosan Body Myopathy, Cardiomyopathy	AR	100	17 of 18
GYS1	Glycogen Storage Disease	AR	99.69	4 of 4
HACD1	Fiber-Type Disproportion Myopathy	-	99.5	-
HNRNPA1	Amyotrophic Lateral Sclerosis, Inclusion Body Myopathy, Paget Disease, Frontotemporal Dementia	AD	99.98	13 of 13
HNRNPA2B1	Inclusion Body Myopathy, Paget Disease, Frontotemporal Dementia	-	99.98	5 of 6
HNRNPDL	Limb-Girdle Muscular Dystrophy	AD	96.58	2 of 2
HRAS	Bladder Cancer, Costello Syndrome, Epidermal Nevus, Giant Pigmented Hairy Nevus, Schimmelpenning-Feuerstein-Mims Syndrome, Thyroid Cancer, Linear Nevus Sebaceus Syndrome	AD	100	34 of 34
HSPB8	Charcot-Marie-Tooth Disease, Neuronopathy	AD	97.59	9 of 9
INPP5K	Muscular Dystrophy, Cataracts, Intellectual Disability, Marinesco-Sjogren Syndrome	AR	92	10 of 10
ISCU	Myopathy	AR	99.94	3 of 3
ITGA7	Muscular Dystrophy, Integrin Alpha-7 Deficiency, Fiber-Type Disproportion Myopathy	AR	99.99	10 of 10
KBTBD13	Nemaline Myopathy	AD	99.66	15 of 15
KLHL40	Nemaline Myopathy	AR	99.98	26 of 26
KLHL41	Nemaline Myopathy	AR	99.92	8 of 8
KLHL9	Distal Myopathy	-	99.97	4 of 4
KY	Myopathy, Kyphoscoliosis, Lateral Tongue Atrophy, Spastic Paraplegia	AR	99.95	3 of 3
LAMA2	Limb Girdle Muscular Dystrophy	AR	100	363 of 377
LAMP2	Danon Disease, Glycogen Storage Disease	X,XD,G	99.96	-
LARGE1	Muscular Dystrophy-Dystroglycanopathy, Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	-
LDB3	Cardiomyopathy, Miofibrillar Myopathy	AD	100	60 of 60
LDHA	Glycogen Storage Disease	AR	99.38	9 of 9
LIMS2	Limb Girdle Muscular Dystrophy	AR	94.23	4 of 4
LMNA	Cardiomyopathy, Charcot-Marie-Tooth Disease, Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Malouf Syndrome, Mandibuloacral Dysplasia, Restrictive Dermopathy, Werner Syndrome, Lipodystrophic Laminopathy, Hypergonadotropic Hypogonadism	AD,AR	100	619 of 620
LMOD3	Nemaline Myopathy	AR	98.68	23 of 26
LPIN1	Rhabdomyolysis	AR	99.98	31 of 31
MAP3K20	Fiber-Type Disproportion Myopathy, Split-Foot Malformation, Mesoaxial Polydactyly	AR	99.68	-
MATR3	Amyotrophic Lateral Sclerosis, Vocal Cord And Pharyngeal Distal Myopathy	AD	99.98	21 of 21
MEGF10	Myopathy, Respiratory Distress, Dysphagia, Areflexia	AR	99.96	20 of 21
MICU1	Myopathy, Extrapyramidal Signs	AR	99.83	7 of 8
MME	Charcot-Marie-Tooth Disease, Spinocerebellar Ataxia, Membranous Nephropathy, Fetomaternal Anti-Neutral Endopeptidase Alloimmunization	AD,AR	100	33 of 33
MMEL1	Primary Biliary Cholangitis	-	100	-

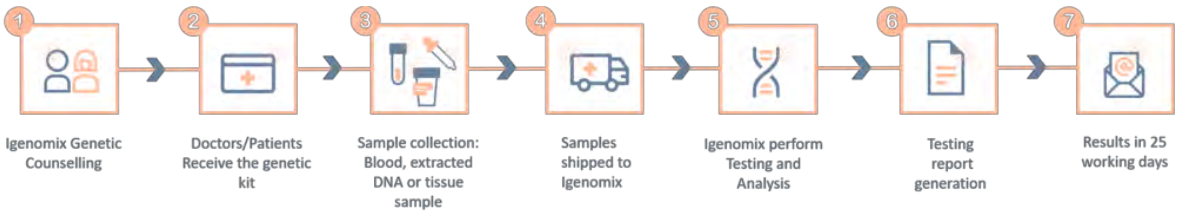
MPDU1	Congenital Disorder Of Glycosylation	AR	100	7 of 7
MSTO1	Myopathy, Ataxia, Pigmentary Retinopathy	AD,AR	88.73	15 of 19
MTM1	Myotubular Myopathy, Centronuclear Myopathy, Abnormal Genitalia	X,XR,G	99.98	-
MTMR14	Centronuclear Myopathy	AD	100	2 of 2
MYBPC1	Arthrogryposis, Contracture Syndrome, Myopathy, Digitotalar Dysmorphism	AD,AR	100	13 of 13
MYBPC3	Cardiomyopathy, Left Ventricular Noncompaction	AD,AR	99.95	1072 of 1079
MYF6	Centronuclear Myopathy	-	100	2 of 2
MYH2	Myopathy	AD,AR	99.98	31 of 31
MYH7	Cardiomyopathy, Ebstein Malformation, Scapuloperoneal Muscular Dystrophy	AD,AR	99.95	1053 of 1054
MYL1	Myopathy	AR	100	2 of 2
MYL2	Cardiomyopathy, Fiber-Type Disproportion Myopathy	AD	100	67 of 67
MYMK	Carey-Fineman-Ziter Syndrome	AR	100	-
MYO18B	Klippel-Feil Syndrome, Myopathy, Facial Dysmorphism	AR	99.39	8 of 9
MYOD1	Myopathy, Diaphragmatic Defects, Respiratory Insufficiency, Dysmorphic Facies, Akinesia Deformation Sequence	AR	99.97	6 of 6
MYOT	Myopathy, Myotilinopathy, Limb-Girdle Muscular Dystrophy	AD	100	17 of 17
MYPN	Cardiomyopathy, Nemaline Myopathy, Cap Myopathy	AD,AR	99.94	49 of 49
NEB	Nemaline Myopathy, Distal Nebulin Myopathy	AR	86.77	304 of 339
ORAI1	Immunodeficiency, Stormorken-Sjaastad-Langslet Syndrome, Tubular Aggregate Myopathy	AD,AR	91.93	20 of 22
PABPN1	Oculopharyngeal Muscular Dystrophy	AD	89.43	0 of 6
PAX7	Myopathy, Scoliosis, Rhabdomyosarcoma	AR	100	17 of 17
PFKM	Glycogen Storage Disease, Muscle Phosphofructokinase Deficiency	AR	99.97	27 of 27
PGAM2	Phosphoglycerate Mutase Deficiency	AR	100	11 of 11
PGK1	Phosphoglycerate Kinase 1 Deficiency, Glycogen Storage Disease	X,XR,G	100	-
PGM1	Congenital Disorder Of Glycosylation	AR	99.96	38 of 40
PHKA1	Muscle Glycogenosis	X,XR,G	99.97	-
PLEC	Epidermolysis Bullosa, Pyloric Atresia, Limb Girdle Muscular Dystrophy, Nail Dystrophy, Aplasia Cutis Congenita	AD,AR	99.98	113 of 113
PNPLA2	Neutral Lipid Storage Disease, Myopathy	AR	100	53 of 53
PNPLA8	Mitochondrial Myopathy, Lactic Acidosis	AR	99.13	6 of 6
POGLUT1	Dowling-Degos Disease, Limb Girdle Muscular Dystrophy	AD,AR	99.94	27 of 27
POLG	Mitochondrial Dna Depletion Syndrome, Ophthalmoplegia, Neuropathy, Dysarthria, Alpers-Huttenlocher Syndrome, Mitochondrial Neurogastrointestinal Encephalomyopathy, Mitochondrial Ataxia Syndrome	AD,AR	99.92	325 of 326
POMGNT1	Muscular Dystrophy-Dystroglycanopathy, Retinitis Pigmentosa, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.91	82 of 83
POMGNT2	Muscular Dystrophy-Dystroglycanopathy, Walker-Warburg Syndrome	AR	100	10 of 10
POMK	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	99.99	8 of 8
POMT1	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	105 of 105
POMT2	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	74 of 74
POPDC3	Limb Girdle Muscular Dystrophy	AR	99.5	3 of 3
PRKAG2	Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff-Parkinson-White Syndrome	AD	99.98	61 of 61

PUS1	Mitochondrial Myopathy, Sideroblastic Anemia	AR	99.58	13 of 14
PYGM	Glycogen Storage Disease, Glycogen Phosphorylase Deficiency	AR	100	167 of 169
PYROXD1	Myopathy	AR	99.92	7 of 8
RBCK1	Polyglucosan Body Myopathy, Immunodeficiency	AR	100	13 of 13
RRM2B	Mitochondrial Dna Depletion Syndrome, Ophthalmoplegia, Kearns-Sayre Syndrome, Neurogastrointestinal Encephalomyopathy	AD,AR	92.38	46 of 46
RXYLT1	Muscular Dystrophy-Dystroglycanopathy, Walker-Warburg Syndrome	AR	99.46	-
RYR1	Central Core Disease Of Muscle, Hyperthermia, Ophthalmoplegia, Fiber-Type Disproportion Myopathy, Centronuclear Myopathy, Multiminicore Disease	AD,AR	97.63	733 of 746
RYR3	Central Core Myopathy, Capillary Malformations, Right Ventricular Dysplasia, Neuroleptic Malignant Syndrome	-	99.98	20 of 20
SCN4A	Hyperkalemic Periodic Paralysis, Hypokalemic Periodic Paralysis, Myasthenic Syndrome, Myotonia, Paramyotonia Congenita Of Von Eulenburg, Acetazolamide-Responsive Myotonia	AD,AR	99.77	136 of 142
SELENON	Fiber-Type Disproportion Myopathy, Rigid Spine Muscular Dystrophy, Multiminicore Myopathy	AD,AR	89	-
SEPTIN9	Neuralgic Amyotrophy	AD	86.94	4 of 4
SGCA	Limb Girdle Muscular Dystrophy	AR	100	119 of 119
SGCB	Limb Girdle Muscular Dystrophy	AR	98.36	55 of 65
SGCD	Cardiomyopathy, Limb Girdle Muscular Dystrophy	AD,AR	99.89	31 of 32
SGCE	Myoclonic Dystonia	AD	99.46	94 of 98
SGCG	Limb Girdle Muscular Dystrophy	AR	100	53 of 55
SIL1	Marinesco-Sjogren Syndrome	AR	100	47 of 48
SLC16A1	Erythrocyte Lactate Transporter Defect, Hyperinsulinemic Hypoglycemia	AD,AR	99.68	12 of 14
SLC22A5	Carnitine Deficiency	AR	100	161 of 162
SLC25A20	Carnitine-Acylcarnitine Translocase Deficiency	AR	100	39 of 39
SMCHD1	Bosma Arhinia Microphthalmia Syndrome, Facioscapulohumeral Muscular Dystrophy, Hyposmia, Nasal And Ocular Hypoplasia, Hypogonadotropic Hypogonadism	AD,MU,D	99.64	131 of 137
SMN1	Spinal Muscular Atrophy	AR	5.2	17 of 91
SMN2	Spinal Muscular Atrophy	AR	7.6	0 of 3
SPEG	Myopathy	AR	99.26	17 of 17
SPTBN4	Myopathy, Neuropathy, Deafness	AR	99.26	10 of 10
SQSTM1	Frontotemporal Dementia, Myopathy, Neurodegeneration, Ataxia, Dystonia, Gaze Palsy, Paget Disease Of Bone, Amyotrophic Lateral Sclerosis	AD,AR	99.25	105 of 107
STAC3	Native American Myopathy	AR	99.98	5 of 5
SUN1	Emery-Dreifuss Muscular Dystrophy, Hyperalphalipoproteinemia, Laminopathy	-	99.78	7 of 7
SUN2	Emery Dreifuss Muscular Dystrophy, Emerinopathy, Laminopathy	-	100	4 of 4
SYNE1	Arthrogryposis, Emery-Dreifuss Muscular Dystrophy, Spinocerebellar Ataxia	AD,AR	99.99	193 of 193
SYNE2	Emery-Dreifuss Muscular Dystrophy	AD	99.94	12 of 12
TCAP	Cardiomyopathy, Limb Girdle Muscular Dystrophy	AD,AR	100	33 of 33
TIA1	Welander Distal Myopathy	AD,AR	100	13 of 13
TK2	Mitochondrial Dna Depletion Syndrome, Ophthalmoplegia	AR	97.08	64 of 65
TMEM126B	Mitochondrial Complex I Deficiency	AR	98.88	4 of 4
TMEM43	Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular Dystrophy	AD	99.98	26 of 26
TNNT1	Nemaline Myopathy	AR	89.94	7 of 8

TNPO3	Limb Girdle Muscular Dystrophy, Biliary Cholangitis	AD	99.98	7 of 7
TOR1AIP1	Limb Girdle Muscular Dystrophy	AR	97.5	5 of 6
TPM2	Arthrogryposis, Fiber-Type Disproportion Myopathy, Nemaline Myopathy, Cap Myopathy, Digitotalar Dysmorphism, Sheldon-Hall Syndrome	AD,AR	100	41 of 41
TPM3	Nemaline Myopathy, Cap Myopathy, Fiber-Type Disproportion Myopathy	AD,AR	100	27 of 27
TRAPPC11	Intellectual Disability, Hyperkinetic Movement, Truncal Ataxia, Limb-Girdle Muscular Dystrophy, Triple A Syndrome	AR	99.97	18 of 18
TRIM32	Bardet-Biedl Syndrome, Limb Girdle Muscular Dystrophy	AR	100	17 of 17
TRIP4	Muscular Dystrophy, Spinal Muscular Atrophy, Respiratory Failure, Skin Abnormalities, Joint Hyperlaxity	AR	99.92	3 of 3
TTN	Cardiomyopathy, Limb Girdle Muscular Dystrophy, Tibial Muscular Dystrophy, Centronuclear And Multiminicore Myopathy	AD,AR	97.93	1153 of 1219
UNC45B	Cataract	AD	99.72	6 of 6
VCP	Amyotrophic Lateral Sclerosis, Frontotemporal Dementia, Charcot-Marie-Tooth Disease, Inclusion Body Myopathy, Paget Disease, Non-Fluent Aphasia, Spastic Paraplegia	AD	100	68 of 69
VMA21	Myopathy	X,XR,G	99.94	-
VPS13A	Choreoacanthocytosis	AR	99.37	120 of 122

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

Methodology



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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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