Arthrogryposis and Congenital Myasthenic Syndrome

Precision Panel

Overview

Arthrogryposis or arthrogryposis multiplex congenita (AMC) is a group of nonprogressive conditions characterized by multiple joint contractures found throughout the body at birth. It usually appears as a feature of other neuromuscular conditions or part of systemic diseases. Primary cases may present prenatally with decreased fetal movements associated with joint contractures as well as brain abnormalities, decreased muscle bulk and polyhydramnios whereas secondary causes may present with isolated contractures. Congenital Myasthenic Syndromes (CMS) are a clinically and genetically heterogeneous group of disorders characterized by impaired neuromuscular transmission. Clinically they usually present with abnormal fatigability upon exertion, transient weakness of extra-ocular, facial, bulbar, truncal or limb muscles. Severity ranges from mild, phasic weakness, to disabling permanent weakness with respiratory difficulties and ultimately death. The mode of inheritance of these diseases typically follows and autosomal recessive pattern, although dominant forms can be seen.

The Igenomix Arthrogryposis and Congenital Myasthenic Syndrome Precision Panel can be as a tool for an accurate diagnosis 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 Arthrogryposis and Congenital Myasthenic Syndrome Precision Panel is used for patients with a clinical suspicion or diagnosis with or without the following symptoms:

- Limb deformities: compression, absent patella, dislocated radial heads etc
- Connective tissue abnormalities: pterygium, shortening, webs etc
- Facial deformities: asymmetry, flat nasal bridge, hemangioma
- Jaw deformities
- Scoliosis
- Facial deformities
- Hernias
- Seizures
- Joint contractures
- Fatigable weakness at birth affecting ocular and other cranial muscles: ocular, bulbar, limb muscles
- Respiratory insufficiency with sudden apnea
- Feeding difficulties

- Positive family history of congenital myasthenic syndrome

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 with acetylcholinesterase inhibitors.
- 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**
ABCC8	Permanent Neonatal Diabetes Mellitus With Or Without Neurologic Features, Dend Syndrome	AD,AR	99.98	710 of 712
ACADM	Medium Chain Acyl-CoA Dehydrogenase Deficiency	AR	99.98	181 of 181
ACTA1	Congenital Myopathy With Fiber-Type Disproportion, Nemaline Myopathy, Zebra Body Myopathy	AD,AR	100	224 of 224
ADCY6	Lethal Congenital Contracture Syndrome, Hypomyelination Neuropathy-Arthrogryposis Syndrome	AR	100	2 of 2
ADGRG6	Lethal Congenital Contracture Syndrome	AR	99.91	NA of NA
AGRN	Congential Myasthenic Syndrome	AR	99.71	18 of 18
AIMP1	Hypomyelinating Leukodystrophy, Autosomal Recessive Non-Syndromic Intellectual Disability	AR	100	10 of 10
AK9	Postsynaptic Congenital Myasthenic Syndromes	-	98.37	4 of 4
ALG14	Congenital Myasthenic Syndrome	AR	99.99	7 of 7
ALG2	Congenital Disorder Of Glycosylation Type II	AR	99.61	7 of 7
ALG3	Congenital Disorder Of Glycosylation Type Id	AR	99.2	25 of 25
ASCC1	Spinal Muscular Atrophy With Congenital Bone Fractures	AR	99.97	6 of 6
ATAD1	Hereditary Hyperekplexia	AR	99.97	3 of 3
AUTS2	Autosomal Dominant Metal Retardation, Autism Spectrum Disorder	AD	99.63	9 of 17
BICD2	Autosomal Dominant Childhood-Onset Proximal Spinal Muscular Atrophy	AD	99.94	39 of 39
BIN1	Autosomal Recessive Centronuclear Myopathy, Autosomal Dominant Centronuclear Myopathy	AR	100	20 of 20
C12ORF65	Combined Oxidative Phosphorylation Deficiency, Autosomal Recessive Spastic Paraplegia	AR	na	na
CACNA1E	Epileptic Encephalopathy	AD	99.94	25 of 25
CASK	Nonspherocytic Hemolytic Anemia, Mental Retardation And Microcephaly With Pontine And Cerebellar Hypoplasia, Early Infantile Epileptic Encephalopathy	X,XR,XD,G	99.98	NA of NA
CCDC47	Trichohepatoneurodevelopmental Syndrome	AR	99.94	5 of 5
CDK5	Lissencephaly With Cerebellar Hypoplasia	AR	100	5 of 5
CEP55	Multinucleated Neurons, Anhydramnios, Renal Dysplasia, Cerebellar Hypoplasia, And Hydranencephaly, Meckel Syndrome	AR	99.22	3 of 3
CFL2	Nemaline Myopathy	AR	99.98	9 of 9
CHAT	Congenital Myasthenic Syndrome Associated With Episodic Apnea	AR	100	49 of 49
CHMP1A	Pontocerebellar Hypoplasia Type 8	AR	100	4 of 4
CHRNA1	Multiple Pterygium Syndrome, Congenital Myasthenic Syndrome	AD,AR	100	35 of 35
CHRNB1	Congential Myasthenic Syndrome	AD,AR	95	9 of 9
CHRND	Multiple Pterygium Syndrome, Congenital Myasthenic Syndrome, Congenital	AD,AR	100	31 of 31

CHRNE	Familial Infantile Myasthenia, Congenital Myasthenic Syndrome	AD,AR	99.87	138 of 138
CHRNG	Multiple Pterygium Syndrome	AR	100	36 of 36
CHST14	Musculocontractural Ehlers-Danlos Syndrome	AR	97.7	21 of 22
CHUK	Cocoon Syndrome	AR	100	5 of 5
CNTNAP1	Lethal Congenital Contracture Syndrome, Congenital Hypomyelinating Neuropathy	AR	99.97	25 of 25
COL13A1	Congenital Myasthenic Syndrome	AR	99.97	16 of 16
COL6A2	Bethlem Myopathy , Congenital Myosclerosis, Ullrich Congenital Muscular Dystrophy	AD,AR	100	223 of 225
COLQ	Endplate Acetylcholinesterase Deficiency, Synaptic Congenital Myasthenic Syndromes	AR	100	70 of 71
DHCR24	Desmosterolosis	AR	100	10 of 10
DOK7	Fetal Akinesia Deformation Sequence, Limb-Girdle Myasthenia, Postsynaptic Congenital Myasthenic Syndromes	AR	99.88	72 of 72
DPAGT1	Congenital Disorder Of Glycosylation, Type Ij, Congenital Myasthenic Syndrome	AR	100	41 of 41
DSE	Musculocontractural Ehlers-Danlos Syndrome	AR	99.94	3 of 3
ECEL1	Distal Arthrogryposis Type 5d	AR	99.52	39 of 39
EGR2	Demyelinating Charcot-Marie-Tooth Disease Type 1d, Hypertrophic Neuropathy Of Dejerine-Sottas, Congenital Hypomyelinating Neuropathy	AD,AR	100	23 of 23
ERBB3	Lethal Congenital Contracture Syndrome	AD,AR	99.91	6 of 6
ERCC1	Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome Type 2	AR	93.12	6 of 6
ERCC2	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum Complementation Group D, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	100	102 of 102
ERCC5	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum Complementation Group G, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	99.94	58 of 58
ERCC6	Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome Type B, De Sanctis-Cacchione Syndrome Type 3	AD,AR	99.98	127 of 128
ERGIC1	Neurogenic Arthrogryposis Multiplex Congenita	AR	100	2 of 2
EXOSC3	Pontocerebellar Hypoplasia Type 1b	AR	100	19 of 20
FAM20C	Lethal Osteosclerotic Bone Dysplasia	AR	97.8	29 of 29
FBN2	Congenital Contractural Arachnodactyly	AD	100	115 of 115
FHL1	Reducing Body Myopathy, Scapuloperoneal Myopathy, Uruguay Faciocardiomusculoskeletal Syndrome, X-linked Emery-Dreifuss Muscular Dystrophy	X,XR,XD,G	99.98	NA of NA
FIG4	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease Type 4j, Cleidocranial Dysplasia With Micrognathia, Absent Thumbs, And Distal , Polymicrogyria, Bilateral Temporooccipital, Yunis-Varon Syndrome	AD,AR	99.92	72 of 72
FKBP10	Bruck Syndrome, Osteogenesis Imperfecta Type XI, Kuskokwim Syndrome	AR	100	51 of 51
FKTN	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Limb-Girdle Muscular Dystrophy Type 2m, Congenital Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	98	54 of 56
FLAD1	Lipid Storage Myopathy Due To Flavin Adenine Dinucleotide Synthetase Deficiency	AR	97.13	13 of 14
FLVCR2	Proliferative Vasculopathy And Hydranencephaly-Hydrocephaly Syndrome	AR	99.97	16 of 16
GBA	Gaucher Disease-Ophthalmoplegia-Cardiovascular Calcification Syndrome, Hereditary Late-Onset Parkinson Disease	AD,AR	100	469 of 471
GBE1	Glycogen Storage Disease IV, Adult Polyglucosan Body Disease	AR	99.95	71 of 74
GCK	Permanent Neonatal Diabetes Mellitus, Familial Hyperinsulinemic Hypoglycemia	AD,AR	100	905 of 909
GFM2	Combined Oxidative Phosphorylation Deficiency Type 39	AR	99.35	5 of 7
GFPT1	Congenital Myasthenic Syndromes With Glycosylation Defect	AR	100	57 of 57
GLDN	Lethal Congenital Contracture Syndrome	AR	98.46	13 of 13
GLE1	Congenital Arthrogryposis With Anterior Horn Cell Disease, Lethal Congenital Contracture Syndrome, Amyotrophic Lateral Sclerosis	AR	100	17 of 17
GLI3	Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Postaxial and Preaxial Polydactyly, Acrocallosal Syndrome	AD,AR	100	231 of 231
GMPPB	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Congential Muscular Dystrophy With Cerebellar	AR	99.95	53 of 53

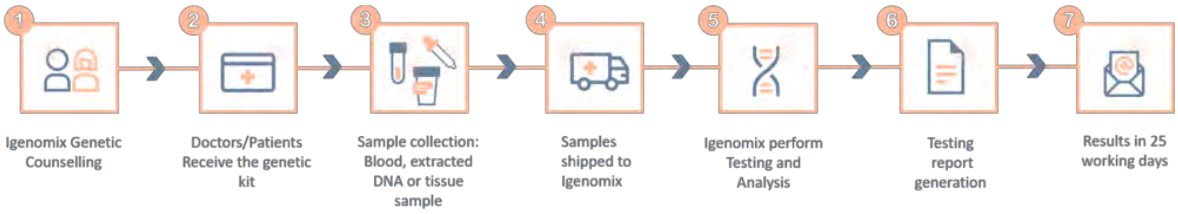
	Involvement, Congenital Myasthenic Syndromes With Glycosylation Defect, Muscle-Eye-Brain Disease			
HSPG2	Dyssegmental Dysplasia, Silverman-Handmaker Type, Schwartz-Jampel Syndrome	AR	99.41	68 of 69
HYMAI	Paternal Uniparental Disomy Of Chromosome 6, Transient Neonatal Diabetes Mellitus	AD	na	na
IBA57	Multiple Mitochondrial Dysfunctions Syndrome, Autosomal Recessive Spastic Paraplegia	AR	93.35	25 of 27
INS	Permanent Neonatal Diabetes Mellitus, Hyperproinsulinemia	AD,AR	100	78 of 84
ITGA6	Epidermolysis Bullosa Junctionalis With Pyloric Atresia, Junctional Epidermolysis Bullosa-Pyloric Atresia Syndrome	AR	100	10 of 10
ITGB4	Epidermolysis Bullosa Junctionalis With Pyloric Atresia, Epidermolysis Bullosa Simplex Weber-Cockayne Type, Aplasia Cutis Congenita	AD,AR	99.12	115 of 115
KAT6B	Genitopatellar Syndrome, Ohdo Syndrome, Blepharophimosis-Intellectual Disability Syndrome	AD	99.97	80 of 80
KBTBD13	Childhood-Onset Nemaline Myopathy	AD	99.66	15 of 15
KCNJ11	Permanent Neonatal Diabetes Mellitus With Or Without Neurologic Features, Hyperinsulinemic Hypoglycemia, Dend Syndrome	AD,AR	100	190 of 191
KIAA1109	Alkuraya-Kucinskas Syndrome	AR	99.95	21 of 21
KIF14	Meckel Syndrome, Autosomal Recessive Primary Microcephaly	AR	99.84	18 of 18
KIF1A	Autosomal Dominant Mental Retardation Neuropathy, Hereditary Sensory And Autonomic Type II, Autosomal Spastic Paraplegia Type 30, Peho Syndrome	AD,AR	100	76 of 76
KIF5C	Cortical Dysplasia, Complex, With Other Brain Malformations	AD	99.96	7 of 7
KLHL40	Severe Congenital Nemaline Myopathy	AR	99.98	26 of 26
KLHL41	Childhood-Onset Nemaline Myopathy	AR	99.92	8 of 8
LAMB2	Pierson Syndrome , Synaptic Congenital Myasthenic Syndromes	AR	100	129 of 129
LGI4	Arthrogryposis Multiplex Congenita, Neurogenic, With Myelin Defect , Hypomyelination Neuropathy-Arthrogryposis Syndrome	AR	99.86	9 of 9
LMNA	Charcot-Marie-Tooth Disease Axonal Type 2b1 , Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Familial Partial Lipodystrophy Type 2, Malouf Syndrome, Mandibuloacral Dysplasia, Congenital Muscular Dystrophy, Atypical Werner Syndrome	AD,AR	100	619 of 620
LMOD3	Severe Congenital Nemaline Myopathy	AR	98.68	23 of 26
LRP4	Cenani-Lenz Syndactyly Syndrome, Congenital Myasthenic Syndrome, Sclerosteosis, Cenani-Lenz Syndrome	AD,AR	100	32 of 32
MAGEL2	Prader-Willi Syndrome	AD	99.99	43 of 48
MED13L	Mental Retardation And Distinctive Facial Features With Or Without Cardiac Defects, Developmental Delay-Facial Dysmorphism Syndrome	AD	100	90 of 92
MPZ	Axonal Type Charcot-Marie-Tooth Disease, Demyelinating Type Charcot-Marie-Tooth Disease, Hypertrophic Neuropathy Of Dejerine-Sottas, Congenital Hypomyelinating Neuropathy, Roussy-Levy Hereditary Areflexic Dystasia, Charcot-Marie-Tooth Disease Type 1b, Roussy-Levy Syndrome	AD,AR	99.98	245 of 245
MTM1	Myotubular Myopathy, X-linked Centronuclear Myopathy , X-linked Myotubular Myopathy-Abnormal Genitalia Syndrome	X,XR,G	99.98	NA of NA
MUSK	Fetal Akinesia Deformation Sequence, Congenital Myasthenic Syndrome	AR	95.58	23 of 25
MYBPC1	Distal Arthrogryposis Type 1b, Lethal Congenital Contracture Syndrome, Congenital Myopathy With Tremor, Digitotalar Dysmorphism	AD,AR	100	13 of 13
MYH2	Proximal Myopathy And Ophthalmoplegia	AD,AR	99.98	31 of 31
MYH3	Distal Arthrogryposis, Contractures, Pterygia, And Spondylocarpostarsal Fusion Syndrome, Autosomal Recessive Multiple Pterygium Syndrome, Digitotalar Dysmorphism, Freeman-Sheldon Syndrome, Sheldon-Hall Syndrome	AD,AR	100	46 of 47
MYH8	Carney Complex Variant, Trismus-Pseudocamptodactyly Syndrome	AD	100	6 of 6
MYO9A	Congenital Myasthenic Syndrome	AR	99.62	7 of 7
MYOD1	Congenital Myopathy With Diaphragmatic Defects, Respiratory Insufficiency, And Dysmorphic Facies, Fetal Akinesia Deformation Sequence	AR	99.97	6 of 6
MYPN	Nemaline Myopathy, Childhood-Onset Nemaline Myopathy	AD,AR	99.94	49 of 49
NALCN	Congenital Contractures Of The Limbs And Face, Hypotonia, And Developmental Delay, Digitotalar Dysmorphism, Freeman-Sheldon Syndrome, Hypotonia-Speech Impairment-Severe Cognitive Delay Syndrome, Sheldon-Hall Syndrome	AD,AR	99.97	69 of 69
NEB	Nemaline Myopathy, Childhood-Onset Nemaline Myopathy, Distal Nebulin Myopathy	AR	86.77	304 of 339

NEK9	Arthrogryposis, Perthes Disease, And Upward Gaze Palsy, Lethal Congenital Contracture Syndrome	AR	99.98	4 of 4
NUP88	Fetal Akinesia Deformation Sequence	AR	95.82	3 of 3
PDX1	Pancreatic Permanent Neonatal Diabetes Mellitus	AD,AR	98.02	32 of 36
PHGDH	Neu-Laxova Syndrome, Phosphoglycerate Dehydrogenase Deficiency	AR	100	26 of 26
PI4KA	Polymicrogyria, Perisylvian, With Cerebellar Hypoplasia And Arthrogryposis	AR	99.76	4 of 4
PIEZO2	Distal Arthrogryposis, Gordon Syndrome, Marden-Walker Syndrome, Arthrogryposis-Oculomotor Limitation-Electroretinal Anomalies Syndrome	AD,AR	96.93	37 of 37
PIGS	Glycosylphosphatidylinositol Biosynthesis Defect	AR	100	6 of 6
PIP5K1C	Lethal Congenital Contracture Syndrome	AR	99.83	3 of 3
PLAGL1	Paternal Uniparental Disomy Of Chromosome 6, Transient Neonatal Diabetes Mellitus	-	95.56	2 of 2
PLEC	Epidermolysis Bullosa Junctionalis With Pyloric Atresia, Epidermolysis Bullosa Simplex	AD,AR	99.98	113 of 113
PLOD2	Bruck Syndrome	AR	99.97	29 of 29
PLXND1	Moebius Syndrome	-	98.44	6 of 6
PMM2	Congenital Disorder Of Glycosylation Type Ia	AR	100	127 of 129
PPP3CA	Arthrogryposis, Cleft Palate, Craniosynostosis, And Impaired Intellectual Development, Undetermined Early-Onset Epileptic Encephalopathy	AD	99.98	16 of 16
PREPL	Congenital Myasthenic Syndrome, 2p21 Microdeletion Syndrome, Hypotonia-Cystinuria Syndrome	AR	99.92	7 of 12
PSAT1	Neu-Laxova Syndrome, Phosphoserine Aminotransferase Deficiency	AR	99.95	9 of 9
PSMB8	Proteasome-Associated Autoinflammatory Syndrome 1 And Digenic Forms	AR	100	11 of 11
RAPSN	Fetal Akinesia Deformation Sequence, Congenital Myasthenic Syndrome	AR	99.98	59 of 61
RARS2	Pontocerebellar Hypoplasia Type 6	AR	99.98	39 of 40
REV3L	Moebius Syndrome		99.08	7 of 7
RFT1	Congenital Disorder Of Glycosylation Type In	AR	99.98	18 of 18
RIPK4	Popliteal Pterygium Syndrome Lethal Type, Bartsocas-Papas Syndrome, Chand Syndrome	AR	99.98	16 of 16
RYR1	Central Core Disease Of Muscle, Minicore Myopathy With External Ophthalmoplegia , Myopathy, Congenital, With Fiber-Type Disproportion, Centronuclear Myopathy, Congenital Multicore Myopathy With External Ophthalmoplegia, Congenital Myopathy With Myasthenic-Like Onset	AD,AR	97.63	733 of 746
SCN4A	Congenital Myasthenic Syndrome, Paramyotonia Congenita Of Von Eulenburg	AD,AR	99.77	136 of 142
SCO2	Autosomal Recessive Axonal Charcot-Marie-Tooth Disease Due To Copper Metabolism Defect, Leigh Syndrome With Cardiomyopathy	AD,AR	100	38 of 38
SELENON	Congoenital Myopathy With Fiber-Type Disproportion, Rigid Spine Muscular Dystrophy, Classic Multiminicore Myopathy	AD,AR	89	NA of NA
SHPK	Isolated Sedoheptulokinase Deficiency	-	99.96	2 of 2
SLC18A3	Congenital Myasthenic Syndrome, Fetal Akinesia Deformation Sequence	AR	99.97	5 of 5
SLC25A1	Congenital Myasthenic Syndrome	AR	90	23 of 25
SLC35A3	Arthrogryposis, Mental Retardation, And Seizures, Autism Spectrum Disorder-Epilepsy-Arthrogryposis Syndrome	AR	99.94	5 of 5
SLC5A7	Congenital Myasthenic Syndrome, Distal Neuronopathy Hereditary Motor Type VIIa	AD,AR	99.92	21 of 21
SLC6A9	Glycine Encephalopathy With Normal Serum Glycine	AR	99.99	5 of 5
SLC9A6	Christianson Syndrome	X,XD,G	98.87	NA of NA
SMN1	Spinal Muscular Atrophy	AR	5.2	17 of 91
SMN2	Spinal Muscular Atrophy	AR	7.6	0 of 3
SNAP25	Congenital Myasthenic Syndromes	AD	100	6 of 6
SOX10	Peripheral Demyelinating Neuropathy, Waardenburg Syndrome	AD	99.74	139 of 147
STAC3	Native American Myopathy	AR	99.98	5 of 5
STAT3	Multisystem Autoimmune Disease, Permanent Neonatal Diabetes Mellitus	AD	100	171 of 171
STIM1	Immune Dysfunction With T-Cell Inactivation Due To Calcium Entry Defect, Myopathy, Tubular Aggregate, Stormorken Syndrome, Stormorken-Sjaastad-Langslet Syndrome, Tubular Aggregate Myopathy	AD,AR	100	28 of 28
SYNE1	Arthrogryposis Multiplex Congenita, Emery-Dreifuss Muscular Dystrophy, Autosomal Recessive Spinocerebellar Ataxia	AD,AR	99.99	193 of 193
SYT2	Congenital Myasthenic Syndrome With Or Without Motorneuropathy	AD	99.98	4 of 4

TBCD	Progressive Encephalopathy, Early-Onset, With Brain Atrophy And Thin Corpus Callosum	AR	94.89	28 of 28
TGFB3	Loeys-Dietz Syndrome, Familial Thoracic Aortic Aneurysm And Aortic Dissection	AD	100	34 of 35
TK2	External Ophthalmoplegia With Mitochondrial DNA Deletions	AR	97.08	64 of 65
TNNI2	Distal Arthrogryposis Type 2b, Digitotalar Dysmorphism, Sheldon-Hall Syndrome	AD	100	11 of 11
TNNT1	Nemaline Myopathy	AR	89.94	7 of 8
TNNT3	Distal Arthrogryposis, Digitotalar Dysmorphism, Sheldon-Hall Syndrome	AD	99.98	5 of 5
TPM2	Distal Arthrogryposis, Congenital Myopathy With Fiber-type Disproportion, Nemaline Myopathy, Cap Myopathy, Digitotalar Dysmorphism, Sheldon-Hall Syndrome	AD,AR	100	41 of 41
TPM3	Congenital Myopathy With Fiber-Type Disproportion, Nemaline Myopathy, Cap Myopathy	AD,AR	100	27 of 27
TRIP4	Congenital Muscular Dystrophy, Spinal Muscular Atrophy With Congenital Bone Fractures, Congenital Muscular Dystrophy-Respiratory-Skin Abnormalities-Joint Hyperlaxity Syndrome	AR	99.92	3 of 3
TRPV4	Brachyrachia, Familial Digital Arthropathy-Brachydactyly, Hereditary Motor And Sensory Neuropathy, Metatropic Dysplasia, Parastremmatic Dwarfism, Scapuloperoneal Spinal Muscular Atrophy, Spondylometaphyseal Dysplasia	AD	100	88 of 88
TSEN2	Pontocerebellar Hypoplasia	AR	95.47	4 of 5
TSEN54	Fatal Infantile Encephalopathy With Olivopontocerebellar Hypoplasia	AR	96.94	20 of 22
UBA1	Infantile-Onset X-linked Spinal Muscular Atrophy	X,XR,G	99.58	NA of NA
VAMP1	Spastic Ataxia, Congenital Myasthenic Syndrome	AD,AR	99.51	8 of 8
VIPAS39	Arthrogryposis, Renal Dysfunction, And Cholestasis	AR	100	15 of 15
VPS33B	Arthrogryposis, Renal Dysfunction, And Cholestasis	AR	100	62 of 62
VRK1	Pontocerebellar Hypoplasia	AR	99.64	15 of 15
YY1	Gabriele-de Vries Syndrome	AD	99.89	13 of 13
ZBTB42	Lethal Congenital Contracture Syndrome	AR	99.81	1 of 1
ZC4H2	Wieacker-Wolff Syndrome, Intellectual Disability-Developmental Delay-Contractures Syndrome	X,XR,XD,G	99.69	NA of NA
ZFP57	Transient Neonatal Diabetes Mellitus	AD	100	15 of 15
ZMPSTE24	Mandibuloacral Dysplasia With Type B Lipodystrophy, Hutchinson-Gilford Progeria Syndrome	AR	100	35 of 36
ZNF335	Primary Autosomal Recessive Microcephaly, Microcephalic Primordial Dwarfism	AR	99.83	20 of 20
ZNHIT3	Peho Syndrome	AR	73.96	1 of 1

*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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