PANEL NEUROMUSCULAR

Nuestro **panel neuromuscular** es ideal para pacientes con enfermedades musculares. Incluye genes que causan enfermedades neurológicas y cubre trastornos, como miopatías metabólicas, distrofias musculares, Charcot-Marie-Tooth, síndromes miasténicos congénitos, miopatías congénitas, miopatías miofibrilares, miopatías nemalínicas y otros síndromes con hipotonía, miotonía o debilidad. La artrogriposis se incluye para el diagnóstico diferencial de los trastornos neuromusculares de aparición temprana. Si existe una alta sospecha diagnóstica de distrofia muscular de Duchenne, recomendamos que el médico ordene un análisis de deleción/duplicación por MLPA dirigido al gen *DMD como un servicio adicional.*

Nº de genes:	354
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	Análisis de CNV incluido Análisis de Expansión de repetición: DMPK

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

Artrogriposis

Miopatía de Bethlem

Enfermedad de Charcot-Marie-Tooth

Síndrome miasténico congénito

Miopatía congénita

Síndrome de Déjerine-Sottas

Hiperekplexia

Hipotonía

Hipertermia maligna

miopatías metabólicas

Distrofia muscular

Distrofia muscular-distroglicanopatía tipo A

miopatía miofibrilar

Síndrome de miopatía-rabdomiolisis

miopatía nemalínica

Miotonía congénita no distrófica

Distrofia muscular de Ullrich

Gene	OMIM (Gene)	Associated diseases (OMIM)	Inheritance
AARS1	601065	Trichothiodystrophy 8, nonphotosensitive;Charcot-Marie-Tooth disease, axonal, type 2N;Developmental and epileptic encephalopathy 29;?Leukoencephalopathy, hereditary diffuse, with spheroids 2	AR, AD
ABHD5	604780	Chanarin-Dorfman syndrome	AR
ACAD9	611103	Mi complex I deficiency, nuclear type 20	AR
ACADM	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of	AR
ACADVL	609575	VLCAD deficiency	AR

ACTA1	102610	Nemaline myopathy 3, AD or recessive;?Myopathy, scapulohumeroperoneal;Myopathy, congenital, with fiber-type disproportion 1;Myopathy, actin, congenital, with cores;Myopathy, actin, congenital, with excess of thin myofilaments	AD, AR, AD
ACTG2	102545	Visceral myopathy;Megacystis-microcolon-intestinal hypoperistalsis syndrome 5	AD
ADGRG6	612243	Lethal congenital contracture syndrome 9	AR
AGL	610860	Glycogen storage disease IIIb;Glycogen storage disease IIIa	AR
AGRN	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	AR
AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	AR
AIFM1	300169	Cowchock syndrome;Combined oxidative phosphorylation deficiency 6;Deafness, XL 5;Spondyloepimetaphyseal dysplasia, XL, with hypomyelinating leukodystrophy	XLR
ALDOA	103850	Glycogen storage disease XII	AR

ALG14	612866	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies;?Myasthenic syndrome, congenital, 15, without tubular aggregates;Myopathy, epilepsy, and progressive cerebral atrophy	AR
ALG2	607905	?Congenital disorder of glycosylation, type Ii;Myasthenic syndrome, congenital, 14, with tubular aggregates	AR
AMPD1	102770	Myopathy due to myoadenylate deaminase deficiency	AR
ANO5	608662	Gnathodiaphyseal dysplasia;Muscular dystrophy, limb-girdle, AR 12;Miyoshi muscular dystrophy 3	AD, AR
ARHGEF9	300429	Developmental and epileptic encephalopathy 8	XL
ASAH1	613468	Spinal muscular atrophy with progressive myoclonic epilepsy;Farber lipogranulomatosis	AR
ASCC1	614215	Spinal muscular atrophy with congenital bone fractures 2;Barrett esophagus/esophageal adenocarcinoma	AR
ATAD1	614452	Hyperekplexia 4	AR

ATL1	606439	Spastic paraplegia 3A, AD;Neuropathy, hereditary sensory, type ID	AD
ATP2A1	108730	Brody myopathy	AR
ATP7A	300011	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3	XLR
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	AR
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	AR
BAG3	603883	Cardiomyopathy, dilated, 1HH;Myopathy, myofibrillar, 6	AD
BICD2	609797	Spinal muscular atrophy, lower extremity-predominant, 2A, AD;Spinal muscular atrophy, lower extremity-predominant, 2B, AD	AD
BIN1	601248	Centronuclear myopathy 2	AR
BSCL2	606158	Lipodystrophy, congenital generalized, type 2;Encephalopathy, progressive, with or without lipodystrophy;Silver spastic paraplegia syndrome;Neuropathy, distal hereditary motor, type VC	AR, AD
BVES	604577	Muscular dystrophy, limb-girdle, AR 25	AR

CACNA1S	114208	Thyrotoxic periodic paralysis, susceptibility to, 1;Malignant hyperthermia susceptibility 5;Malignant hyperthermia, susceptibility to, 5;Hypokalemic periodic paralysis, type 1	AD
CAPN3	114240	Muscular dystrophy, limb-girdle, AD 4;Muscular dystrophy, limb-girdle, AR 1	AD, AR
CASK	300172	Mental retardation, with or without nystagmus;Mental retardation and microcephaly with pontine and cerebellar hypoplasia;FG syndrome 4	XLD
CASQ1	114250	Myopathy, vacuolar, with CASQ1 aggregates	AD
CAV1	601047	Pulmonary hypertension, primary, 3;Lipodystrophy, familial partial, type 7;?Lipodystrophy, congenital generalized, type 3	AD, AR
CAV3	601253	Rippling muscle disease 2;Cardiomyopathy, familial hypertrophic;Creatine phosphokinase, elevated serum;Long QT syndrome 9;Myopathy, distal, Tateyama type	AD, AD, DD
CCDC78	614666	?Centronuclear myopathy 4	AD
CFL2	601443	Nemaline myopathy 7, AR	AR
CHAT	118490	Myasthenic syndrome, congenital, 6, presynaptic	AR

CHCHD10	615903	?Myopathy, isolated Mi, AD;Spinal muscular atrophy, Jokela type;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2	AD
CHKB	612395	Muscular dystrophy, congenital, megaconial type	AR
CHRNA1	100690	Myasthenic syndrome, congenital, 1B, fast-channel;Myasthenic syndrome, congenital, 1A, slow-channel;Multiple pterygium syndrome, lethal type	AD, AR, AD, AR
CHRNB1	100710	Myasthenic syndrome, congenital, 2A, slow-channel;?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	AD, AR
CHRND	100720	?Myasthenic syndrome, congenital, 3A, slow-channel;Myasthenic syndrome, congenital, 3B, fast-channel;Multiple pterygium syndrome, lethal type;?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	AD, AR

CHRNE	100725	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency;Myasthenic syndrome, congenital, 4A, slow-channel;Myasthenic syndrome, congenital, 4B, fast-channel	AR, AD, AR
CHRNG	100730	Multiple pterygium syndrome, lethal type;Escobar syndrome	AR
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural type 1	AR
CLCN1	118425	Myotonia congenita, dominant;Myotonia congenita, recessive	AD, AR
CNTNAP1	602346	Lethal congenital contracture syndrome 7;Hypomyelinating neuropathy, congenital, 3	AR
COL12A1	120320	?Ullrich congenital muscular dystrophy 2;Bethlem myopathy 2	AR, AD
COL13A1	120350	Myasthenic syndrome, congenital, 19	AR
COL6A1	120220	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1	AD, AR
COL6A2	120240	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;?Myosclerosis, congenital	AD, AR, AR
COL6A3	120250	Dystonia 27;Ullrich congenital muscular dystrophy 1;Bethlem myopathy 1	AR, AD, AR

COLQ	603033	Myasthenic syndrome, congenital, 5	AR
COQ2	609825	Coenzyme Q10 deficiency, primary, 1;Multiple system atrophy, susceptibility to	AR, AD, AR
COX6A1	602072	Charcot-Marie-Tooth disease, recessive intermediate D	AR
CPT2	600650	CPT II deficiency, myopathic, stress-induced;CPT II deficiency, infantile;Encephalopathy, acute, infection-induced, 4, susceptibility to;CPT II deficiency, lethal neonatal	AD, AR, AR
CRLF1	604237	Cold-induced sweating syndrome 1	AR
CRPPA	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	AR
CRYAB	123590	Cataract 16, multiple types;Cardiomyopathy, dilated, 1II;Myopathy, myofibrillar, 2;Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related	AD, AR, AD, AR
CSRP3	600824	?Cardiomyopathy, dilated, 1M;Cardiomyopathy, hypertrophic, 12	AD
CTDP1	604927	Congenital cataracts, facial dysmorphism, and neuropathy	AR

DAG1	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9	AR
DCTN1	601143	Perry syndrome;Neuronopathy, distal hereditary motor, type VIIB;Amyotrophic lateral sclerosis, susceptibility to	AD, AD, AR
DES	125660	Cardiomyopathy, dilated, 1I;Myopathy, myofibrillar, 1;Scapuloperoneal syndrome, neurogenic, Kaeser type	AD, AD, AR
DGUOK	601465	Portal hypertension, noncirrhotic;Progressive external ophthalmoplegia with Mi DNA deletions, AR 4;Mi DNA depletion syndrome 3 (hepatocerebral type)	AR
DHCR24	606418	Desmosterolosis	AR
DHTKD1	614984	2-aminoadipic 2-oxoadipic aciduria;?Charcot-Marie-Tooth disease, axonal, type 2Q	AR, AD
DMD	300377	Cardiomyopathy, dilated, 3B;Duchenne muscular dystrophy;Becker muscular dystrophy	XL, XLR
DMPK	605377	Myotonic dystrophy 1	AD

DNA2	601810	?Seckel syndrome 8;Progressive external ophthalmoplegia with Mi DNA deletions, AD 6	AR, AD
DNAJB2	604139	Spinal muscular atrophy, distal, AR, 5	AR
DNAJB6	611332	Muscular dystrophy, limb-girdle, AD 1	AD
DNM2	602378	Lethal congenital contracture syndrome 5;Charcot-Marie-Tooth disease, axonal type 2M;Charcot-Marie-Tooth disease, dominant intermediate B;Centronuclear myopathy 1	AR, AD
DNMT1	126375	Neuropathy, hereditary sensory, type IE;Cerebellar ataxia, deafness, and narcolepsy, AD	AD
DOK7	610285	Fetal akinesia deformation sequence 3;Myasthenic syndrome, congenital, 10	AR
DPAGT1	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates;Congenital disorder of glycosylation, type Ij	AR
DPM1	603503	Congenital disorder of glycosylation, type Ie	AR
DPM2	603564	Congenital disorder of glycosylation, type Iu	AR

DPM3	605951	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15;?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15	AR
DYNC1H1	600112	Spinal muscular atrophy, lower extremity-predominant 1, AD;Charcot-Marie-Tooth disease, axonal, type 20;Mental retardation, AD 13	AD
DYSF	603009	Muscular dystrophy, limb-girdle, AR 2;Myopathy, distal, with anterior tibial onset;Miyoshi muscular dystrophy 1	AR
ECEL1	605896	Arthrogryposis, distal, type 5D	AR
EGR2	129010	Dejerine-Sottas disease;Hypomyelinating neuropathy, congenital, 1;Charcot-Marie-Tooth disease, type 1D	AD, AR, AD
ELP1	603722	Dysautonomia, familial;Medulloblastoma	AR, AD, AR, SM
EMD	300384	Emery-Dreifuss muscular dystrophy 1, XL	XLR
ENO3	131370	?Glycogen storage disease XIII	AR

ERCC5	133530	Xeroderma pigmentosum, group G;Cerebrooculofacioskeletal syndrome 3;Xeroderma pigmentosum, group G/Cockayne syndrome	AR
ERCC6	609413	Lung cancer, susceptibility to;UV-sensitive syndrome 1;Premature ovarian failure 11;Macular degeneration, age-related, susceptibility to, 5;Cockayne syndrome, type B;De Sanctis-Cacchione syndrome;Cerebrooculofacioskeletal syndrome 1	AD, SM, AR, AD
ETFA	608053	Glutaric acidemia IIA	AR
ETFB	130410	Glutaric acidemia IIB	AR
ETFDH	231675	Glutaric acidemia IIC	AR
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	AR
EXOSC8	606019	Pontocerebellar hypoplasia, type 1C	AR
FBLN5	604580	Cutis laxa, AR, type IA;Neuropathy, hereditary, with or without age-related macular degeneration;Macular degeneration, age-related, 3;?Cutis laxa, AD 2	AR, AD
FBN2	612570	Contractural arachnodactyly, congenital;Macular degeneration, early-onset	AD
FBXO38	608533	Neuronopathy, distal hereditary motor, type IID	AD

FDX2	614585	Mi myopathy, episodic, with optic atrophy and reversible leukoencephalopathy	AR
FGD4	611104	Charcot-Marie-Tooth disease, type 4H	AR
FHL1	300163	Reducing body myopathy, XL 1b, with late childhood or adult onset;Scapuloperoneal myopathy, XLD;?Uruguay faciocardiomusculoskeletal syndrome;Myopathy, XL, with postural muscle atrophy;Reducing body myopathy, XL 1a, severe, infantile or early childhood onset;Emery-Dreifuss muscular dystrophy 6, XL	XL, XLD, XLR
FIG4	609390	Amyotrophic lateral sclerosis 11;Yunis-Varon syndrome;Charcot-Marie-Tooth disease, type 4J;?Polymicrogyria, bilateral temporooccipital	AD, AR
FKBP10	607063	Osteogenesis imperfecta, type XI;Bruck syndrome 1	AR
FKBP14	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	AR

FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	AR
FKTN	607440	Cardiomyopathy, dilated, 1X;Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	AR
FLAD1	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	AR
FLNC	102565	Cardiomyopathy, familial restrictive 5;Myopathy, myofibrillar, 5;Myopathy, distal, 4;Cardiomyopathy, familial hypertrophic, 26	AD
GAA	606800	Glycogen storage disease II	AR
GAN	605379	Giant axonal neuropathy-1	AR

GARS1	600287	Spinal muscular atrophy, infantile, James type;Charcot-Marie-Tooth disease, type 2D;Neuronopathy, distal hereditary motor, type VA	AD
GBA	606463	Lewy body dementia, susceptibility to;Gaucher disease, type IIIC;Parkinson disease, late-onset, susceptibility to;Gaucher disease, type II;Gaucher disease, type III;Gaucher disease, perinatal lethal;Gaucher disease, type I	AD, AR, AD, mi
GBE1	607839	Glycogen storage disease IV;Polyglucosan body disease, adult form	AR
GDAP1	606598	Charcot-Marie-Tooth disease, axonal, type 2K;Charcot-Marie-Tooth disease, type 4A;Charcot-Marie-Tooth disease, axonal, with vocal cord paresis;Charcot-Marie-Tooth disease, recessive intermediate, A	AD, AR, AR
GFPT1	138292	Myasthenia, congenital, 12, with tubular aggregates	AR
GJB1	304040	Charcot-Marie-Tooth neuropathy, XLD, 1	XLD
GLDN	608603	Lethal congenital contracture syndrome 11	AR
GLE1	603371	Congenital arthrogryposis with anterior horn cell disease;Lethal congenital contracture syndrome 1	AR

GLRA1	138491	Hyperekplexia 1	AD, AR
GLRB	138492	Hyperekplexia 2	AR
GMPPB	615320	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	AR
GNB4	610863	Charcot-Marie-Tooth disease, dominant intermediate F	AD
GNE	603824	Nonaka myopathy;Sialuria	AR, AD
GPHN	603930	Molybdenum cofactor deficiency C	AR
GYG1	603942	Polyglucosan body myopathy 2;?Glycogen storage disease XV	AR
GYS1	138570	Glycogen storage disease 0, muscle	AR
HADHA	600890	HELLP syndrome, maternal, of pregnancy;LCHAD deficiency;Fatty liver, acute, of pregnancy;Mi trifunctional protein deficiency	AR
HADHB	143450	Trifunctional protein deficiency	AR
HINT1	601314	Neuromyotonia and axonal neuropathy, AR	AR

HK1	142600	Retinitis pigmentosa 79;Hemolytic anemia due to hexokinase deficiency;Neurodevelopmental disorder with visual defects and brain anomalies;Neuropathy, hereditary motor and sensory, Russe type	AD, AR
HNRNPDL	607137	Muscular dystrophy, limb-girdle, AD 3	AD
HOXD10	142984	Vertical talus, congenital;Charcot-Marie-Tooth disease, foot deformity of	AD
HRAS	190020	Thyroid carcinoma, follicular, somatic;Spitz nevus or nevus spilus, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Bladder cancer, somatic;Costello syndrome;Nevus sebaceous or woolly hair nevus, somatic;Congenital myopathy with excess of muscle spindles	AD
HSPB1	602195	Neuronopathy, distal hereditary motor, type IIB;Charcot-Marie-Tooth disease, axonal, type 2F	AD
HSPB3	604624	?Neuronopathy, distal hereditary motor, type IIC	AD
HSPB8	608014	Neuronopathy, distal hereditary motor, type IIA;Charcot-Marie-Tooth disease, axonal, type 2L	AD

HSPG2	142461	Schwartz-Jampel syndrome, type 1;Dyssegmental dysplasia, Silverman-Handmaker type	AR
IGHMBP2	600502	Neuronopathy, distal hereditary motor, type VI;Charcot-Marie-Tooth disease, axonal, type 2S	AR
INF2	610982	Glomerulosclerosis, focal segmental, 5;Charcot-Marie-Tooth disease, dominant intermediate E	AD
ISCU	611911	Myopathy with lactic acidosis, hereditary	AR
ITGA7	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency	AR
KARS1	601421	?Charcot-Marie-Tooth disease, recessive intermediate, B;Deafness, congenital, and adult-onset progressive leukoencephalopathy;Leukoencephalopathy, progressive, infantile-onset, with or without deafness;Deafness, AR 89	AR
KAT6B	605880	SBBYSS syndrome;Genitopatellar syndrome	AD
KBTBD13	613727	Nemaline myopathy 6, AD	AD
KCNA1	176260	Episodic ataxia/myokymia syndrome	AD
KCNE3	604433	?Brugada syndrome 6	

KCNJ2	600681	Short QT syndrome 3;Atrial fibrillation, familial, 9;Andersen syndrome	AD
KIF1A	601255	NESCAV syndrome;Spastic paraplegia 30, AR;Neuropathy, hereditary sensory, type IIC;Spastic paraplegia 30, AD	AD, AD, AR, AR
KIF1B	605995	?Charcot-Marie-Tooth disease, type 2A1;Pheochromocytoma;Neuroblastoma, susceptibility to, 1	AD, AD, SM
KIF5A	602821	Amyotrophic lateral sclerosis, susceptibility to, 25;Myoclonus, intractable, neonatal;Spastic paraplegia 10, AD	AD
KLHL40	615340	Nemaline myopathy 8, AR	AR
KLHL41	607701	Nemaline myopathy 9	AR
KLHL7	611119	PERCHING syndrome;Retinitis pigmentosa 42	AR, AD
KY	605739	Myopathy, myofibrillar, 7	AR
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient;Muscular dystrophy, limb-girdle, AR 23	AR
LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities;Pierson syndrome	AR
LAMP2	309060	Danon disease	XLD

LARGE1	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	AR
LDB3	605906	Cardiomyopathy, hypertrophic, 24;Myopathy, myofibrillar, 4;Cardiomyopathy, dilated, 1C, with or without LVNC;Left ventricular noncompaction 3	AD
LDHA	150000	Glycogen storage disease XI	AR
LGI4	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect	AR
LIMS2	607908	?Muscular dystrophy, AR, with cardiomyopathy and triangular tongue	AR
LITAF	603795	Charcot-Marie-Tooth disease, type 1C	AD

LMNA	150330	Malouf syndrome;Emery-Dreifuss muscular dystrophy 3, AR;Emery-Dreifuss muscular dystrophy 2, AD;Hutchinson-Gilford progeria;Muscular dystrophy, congenital;Restrictive dermopathy, lethal;Lipodystrophy, familial partial, type 2;Charcot-Marie-Tooth disease, type 2B1;Mandibuloacral dysplasia;Cardiomyopathy, dilated, 1A;Heart-hand syndrome, Slovenian type	AD, AR
LMOD3	616112	Nemaline myopathy 10	AR
LPIN1	605518	Myoglobinuria, acute recurrent, AR	AR
LRP4	604270	?Myasthenic syndrome, congenital, 17;Cenani-Lenz syndactyly syndrome;Sclerosteosis 2	AR, AD, AR
LRSAM1	610933	Charcot-Marie-Tooth disease, axonal, type 2P	AD, AR
MAGEL2	605283	Schaaf-Yang syndrome	AD
MAMLD1	300120	Hypospadias 2, XL	XLR
MARS1	156560	Charcot-Marie-Tooth disease, axonal, type 2U;?Trichothiodystrophy 9, nonphotosensitive;Interstitial lung and liver disease	AD, AR
MATR3	164015	Amyotrophic lateral sclerosis 21	AD

MED25	610197	Basel-Vanagait-Smirin-Yosef syndrome	AR
MEGF10	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset;Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant	AR
MFN2	608507	Hereditary motor and sensory neuropathy VIA;Charcot-Marie-Tooth disease, axonal, type 2A2B;Charcot-Marie-Tooth disease, axonal, type 2A2A	AD, AR
MICU1	605084	Myopathy with extrapyramidal signs	AR
MPV17	137960	Charcot-Marie-Tooth disease, axonal, type 2EE;Mi DNA depletion syndrome 6 (hepatocerebral type)	AR
MPZ	159440	Charcot-Marie-Tooth disease, type 2I;Dejerine-Sottas disease;Hypomyelinating neuropathy, congenital, 2;Charcot-Marie-Tooth disease, type 2J;Charcot-Marie-Tooth disease, dominant intermediate D;Charcot-Marie-Tooth disease, type 1B;Roussy-Levy syndrome	AD, AD, AR
MT-ATP6			
MT-ATP8			
MT-CO1			
MT-CO2			

MT-CO3			
MT-CYB			
MT-ND1			
MT-ND2			
MT-ND3			
MT-ND4			
MT-ND4L			
MT-ND5			
MT-ND6			
MT-RNR1			
MT-RNR2			
MT-TA			
MT-TC			
MT-TD			
MT-TE			
MT-TF			
MT-TG			
MT-TH			
MT-TI			
MT-TK			
MT-TL1			
MT-TL2			
MT-TM			
MT-TN			
MT-TP			
MT-TQ			
MT-TR			
MT-TS1			
MT-TS2			
MT-TT			
MT-TV			
MT-TW			
MT-TY			
MTM1	300415	Myotubular myopathy, XL	XLR

MTMR14	611089	Centronuclear myopathy, autosomal, modifier of	AD
MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1	AR
MUSK	601296	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency;Fetal akinesia deformation sequence 1	AR
MYBPC1	160794	Arthrogryposis, distal, type 1B;Lethal congenital contracture syndrome 4;Myopathy, congenital, with tremor	AD, AR
MYBPC3	600958	Cardiomyopathy, hypertrophic, 4;Cardiomyopathy, dilated, 1MM;Left ventricular noncompaction 10	AD, AR, AD
MYH2	160740	Proximal myopathy and ophthalmoplegia	AD, AR
MYH3	160720	Arthrogryposis, distal, type 2B3 (Sheldon-Hall);Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B;Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A;Arthrogryposis, distal, type 2A (Freeman-Sheldon)	AD, AR

MYH7	160760	Myopathy, myosin storage, AR;Scapuloperoneal syndrome, myopathic type;Left ventricular noncompaction 5;Cardiomyopathy, dilated, 1S;Cardiomyopathy, hypertrophic, 1;Myopathy, myosin storage, AD;Laing distal myopathy	AR, AD, AD, DD
MYH8	160741	Trismus-pseudocamptodactyly syndrome;Carney complex variant	AD
MYL1	160780	Myopathy, congenital, with fast-twitch (type II) fiber atrophy	AR
MYL2	160781	Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy;Cardiomyopathy, hypertrophic, 10	AR, AD
MYMK	615345	Carey-Fineman-Ziter syndrome	AR
MYO18B	607295	Klippel-Feil syndrome 4, AR, with myopathy and facial dysmorphism	AR
MYO9A	604875	Myasthenic syndrome, congenital, 24, presynaptic	AR
MYOT	604103	Myopathy, myofibrillar, 3;Myopathy, spheroid body	AD
MYPN	608517	Cardiomyopathy, hypertrophic, 22;Cardiomyopathy, dilated, 1KK;Cardiomyopathy, familial restrictive, 4;Nemaline myopathy 11, AR	AD, AR

NALCN	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	AD, AR
NDRG1	605262	Charcot-Marie-Tooth disease, type 4D	AR
NEB	161650	Nemaline myopathy 2, AR;Arthrogryposis multiplex congenita 6	AR
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	AR
OPA1	605290	Optic atrophy 1;?Mi DNA depletion syndrome 14 (encephalocardiomyopathic type);Glaucoma, normal tension, susceptibility to;Optic atrophy plus syndrome;Behr syndrome	AD, AR
OPA3	606580	3-methylglutaconic aciduria, type III;Optic atrophy 3 with cataract	AR, AD
PAX7	167410	Myopathy, congenital, progressive, with scoliosis;Rhabdomyosarcoma 2, alveolar	AR, SM
PDK3	300906	?Charcot-Marie-Tooth disease, XLD, 6	XLD
PFKM	610681	Glycogen storage disease VII	AR
PGAM2	612931	Glycogen storage disease X	AR
PGK1	311800	Phosphoglycerate kinase 1 deficiency	XLR

PGM1	171900	Congenital disorder of glycosylation, type It	AR
PHKA1	311870	Muscle glycogenosis	XLR
PIEZO2	613629	Arthrogryposis, distal, type 3;?Marden-Walker syndrome;Arthrogryposis, distal, type 5;Arthrogryposis, distal, with impaired proprioception and touch	AD, AR
PIP5K1C	606102	Lethal congenital contractural syndrome 3	AR
PLEC	601282	Epidermolysis bullosa simplex, Ogna type;Epidermolysis bullosa simplex with muscular dystrophy;?Epidermolysis bullosa simplex with nail dystrophy;Epidermolysis bullosa simplex with pyloric atresia;Muscular dystrophy, limb-girdle, AR 17	AD, AR
PLEKHG5	611101	Charcot-Marie-Tooth disease, recessive intermediate C;Spinal muscular atrophy, distal, AR, 4	AR
PLOD2	601865	Bruck syndrome 2	AR
PMM2	601785	Congenital disorder of glycosylation, type Ia	AR

PMP22	601097	Neuropathy, recurrent, with pressure palsies;Dejerine-Sottas disease;Roussy-Levy syndrome;?Neuropathy, inflammatory demyelinating;Charcot-Marie-Tooth disease, type 1E;Charcot-Marie-Tooth disease, type 1A	AD, AD, AR, ?AD
PNPLA2	609059	Neutral lipid storage disease with myopathy	AR
POGLUT1	615618	Dowling-Degos disease 4;?Muscular dystrophy, limb-girdle, AR 21	AD, AR
POLG	174763	Progressive external ophthalmoplegia, AR 1;Progressive external ophthalmoplegia, AD 1;Mi recessive ataxia syndrome (includes SANDO and SCAE);Mi DNA depletion syndrome 4B (MNGIE type);Mi DNA depletion syndrome 4A (Alpers type)	AR, AD
POLG2	604983	Mi DNA depletion syndrome 16 (hepatic type);?Mi DNA depletion syndrome 16B (neuroophthalmic type);Progressive external ophthalmoplegia with Mi DNA deletions, AD 4	AR, AD

POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3;Retinitis pigmentosa 76	AR
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8	AR
POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12;?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12	AR
POMT1	607423	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	AR

POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	AR
PREPL	609557	Myasthenic syndrome, congenital, 22	AR
PRKAG2	602743	Glycogen storage disease of heart, lethal congenital;Cardiomyopathy, hypertrophic 6;Wolff-Parkinson-White syndrome	AD
PRPS1	311850	Charcot-Marie-Tooth disease, XLR, 5;Deafness, XL 1;Phosphoribosylpyrophosphate synthetase superactivity;Arts syndrome;Gout, PRPS-related	XLR, XL
PRX	605725	Dejerine-Sottas disease;Charcot-Marie-Tooth disease, type 4F	AD, AR, AR
PYGM	608455	McArdle disease	AR
PYROXD1	617220	Myopathy, myofibrillar, 8	AR
QARS1	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	AR

RAB7A	602298	Charcot-Marie-Tooth disease, type 2B	AD
RAPSN	601592	Fetal akinesia deformation sequence 2;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	AR
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency	AR
REEP1	609139	Spastic paraplegia 31, AD;?Neuronopathy, distal hereditary motor, type VB	AD
RETREG1	613114	Neuropathy, hereditary sensory and autonomic, type IIB	AR
RRM2B	604712	Mi DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy);Progressive external ophthalmoplegia with Mi DNA deletions, AD 5;Mi DNA depletion syndrome 8B (MNGIE type)	AR, AD
RXYLT1	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	AR
RYR1	180901	Malignant hyperthermia susceptibility 1;King-Denborough syndrome;Central core disease;Neuromuscular disease, congenital, with uniform type 1 fiber;Minicore myopathy with external ophthalmoplegia	AD, AD, AR, AR

SBF1	603560	Charcot-Marie-Tooth disease, type 4B3	AR
SBF2	607697	Charcot-Marie-Tooth disease, type 4B2	AR
SCN4A	603967	Paramyotonia congenita;Myotonia congenita, atypical, acetazolamide-responsive;Myasthenic syndrome, congenital, 16;Hyperkalemic periodic paralysis, type 2;Hypokalemic periodic paralysis, type 2	AD, AR
SELENON	606210	Muscular dystrophy, rigid spine, 1;Myopathy, congenital, with fiber-type disproportion	AR, AD, AR
SGCA	600119	Muscular dystrophy, limb-girdle, AR 3	AR
SGCB	600900	Muscular dystrophy, limb-girdle, AR 4	AR
SGCD	601411	Muscular dystrophy, limb-girdle, AR 6;Cardiomyopathy, dilated, 1L	AR
SGCE	604149	Dystonia-11, myoclonic	AD
SGCG	608896	Muscular dystrophy, limb-girdle, AR 5	AR
SH3TC2	608206	Mononeuropathy of the median nerve, mild;Charcot-Marie-Tooth disease, type 4C	AD, AR
SIL1	608005	Marinesco-Sjogren syndrome	AR

SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy	AR
SLC16A1	600682	Hyperinsulinemic hypoglycemia, familial, 7;Erythrocyte lactate transporter defect;Monocarboxylate transporter 1 deficiency	AD, AD, AR
SLC18A3	600336	Myasthenic syndrome, congenital, 21, presynaptic	AR
SLC22A5	603377	Carnitine deficiency, systemic primary	AR
SLC25A1	190315	Myasthenic syndrome, congenital, 23, presynaptic;Combined D-2- and L-2-hydroxyglutaric aciduria	AR
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	AR
SLC25A46	610826	Neuropathy, hereditary motor and sensory, type VIB;Pontocerebellar hypoplasia, type 1E	AR
SLC52A2	607882	Brown-Vialetto-Van Laere syndrome 2	AR
SLC52A3	613350	?Fazio-Londe disease;Brown-Vialetto-Van Laere syndrome 1	AR
SLC5A7	608761	Neuronopathy, distal hereditary motor, type VIIA;Myasthenic syndrome, congenital, 20, presynaptic	AD, AR
SLC6A5	604159	Hyperekplexia 3	AD, AR

SMCHD1	614982	Bosma arhinia microphthalmia syndrome;Fascioscapulohumeral muscular dystrophy 2, digenic	AD, DD
SMN1	600354	Spinal muscular atrophy-3;Spinal muscular atrophy-1;Spinal muscular atrophy-2;Spinal muscular atrophy-4	AR
SMN2	601627	Spinal muscular atrophy, type III, modifier of	AR
SMPD4	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies	AR
SPEG	615950	Centronuclear myopathy 5	AR
SPG11	610844	Spastic paraplegia 11, AR;Amyotrophic lateral sclerosis 5, juvenile;Charcot-Marie-Tooth disease, axonal, type 2X	AR
SPTBN4	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	AR
SPTLC1	605712	Neuropathy, hereditary sensory and autonomic, type IA	AD
SPTLC2	605713	Neuropathy, hereditary sensory and autonomic, type IC	AD
STAC3	615521	Myopathy, congenital, Baily-Bloch	AR
STIM1	605921	Myopathy, tubular aggregate, 1;Stormorken syndrome;Immunodeficiency 10	AD, AR

SUCLA2	603921	Mi DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	AR
SUN2	613569		
SYNE1	608441	Arthrogryposis multiplex congenita 3, myogenic type;Emery-Dreifuss muscular dystrophy 4, AD;Spinocerebellar ataxia, AR 8	AR, AD
SYNE2	608442	Emery-Dreifuss muscular dystrophy 5, AD	AD
SYT2	600104	Myasthenic syndrome, congenital, 7B, presynaptic, AR;Myasthenic syndrome, congenital, 7, presynaptic	AR, AD
TAFAZZIN	300394	Barth syndrome	XLR
TANGO2	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	AR
TBCK	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	AR
TCAP	604488	Muscular dystrophy, limb-girdle, AR 7;Cardiomyopathy, hypertrophic, 25	AR, AD
TFG	602498	Hereditary motor and sensory neuropathy, Okinawa type;?Spastic paraplegia 57, AR	AD, AR

TGFB3	190230	Loeys-Dietz syndrome 5;Arrhythmogenic right ventricular dysplasia 1	AD
TIA1	603518	Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia;Welander distal myopathy	AD, AD, AR
TK2	188250	Mi DNA depletion syndrome 2 (myopathic type);?Progressive external ophthalmoplegia with Mi DNA deletions, AR 3	AR
TMEM43	612048	Arrhythmogenic right ventricular dysplasia 5;Emery-Dreifuss muscular dystrophy 7, AD	AD
TNNI2	191043	Arthrogryposis, distal, type 2B1	AD
TNNT1	191041	Nemaline myopathy 5, Amish type	AR
TNNT3	600692	Arthrogryposis, distal, type 2B2	AD
TNPO3	610032	Muscular dystrophy, limb-girdle, AD 2	AD
TOR1A	605204	Dystonia-1, torsion;Arthrogryposis multiplex congenita 5	AD, AR
TOR1AIP1	614512	?Muscular dystrophy, AR, with rigid spine and distal joint contractures	AR
TPM2	190990	Arthrogryposis, distal, type 2B4;Arthrogryposis, distal, type 1A;CAP myopathy 2;Nemaline myopathy 4, AD	AD

TPM3	191030	Myopathy, congenital, with fiber-type disproportion;Nemaline myopathy 1, AD or recessive;CAP myopathy 1	AD, AR
TRAPPC11	614138	Muscular dystrophy, limb-girdle, AR 18	AR
TRIM2	614141	Charcot-Marie-Tooth disease, type 2R	AR
TRIM32	602290	Muscular dystrophy, limb-girdle, AR 8;?Bardet-Biedl syndrome 11	AR
TRIP4	604501	Spinal muscular atrophy with congenital bone fractures 1;?Muscular dystrophy, congenital, Davignon-Chauveau type	AR
TRPV4	605427	SED, Maroteaux type;Spondylometaphyseal dysplasia, Kozlowski type;Metatropic dysplasia;Brachyolmia type 3;Neuronopathy, distal hereditary motor, type VIII;[Sodium serum level QTL 1];?Avascular necrosis of femoral head, primary, 2;Scapuloperoneal spinal muscular atrophy;Parastremmatic dwarfism;Hereditary motor and sensory neuropathy, type IIc;Digital arthropathy-brachydactyly, familial	AD
TSEN2	608753	Pontocerebellar hypoplasia type 2B	AR

TSFM	604723	Combined oxidative phosphorylation deficiency 3	AR
TTN	188840	Cardiomyopathy, dilated, 1G;Muscular dystrophy, limb-girdle, AR 10;Tibial muscular dystrophy, tardive;Myopathy, myofibrillar, 9, with early respiratory failure;Salih myopathy;Cardiomyopathy, familial hypertrophic, 9	AR, AD
TWNK	606075	Progressive external ophthalmoplegia with Mi DNA deletions, AD 3;Perrault syndrome 5;Mi DNA depletion syndrome 7 (hepatocerebral type)	AD, AR
TYMP	131222	Mi DNA depletion syndrome 1 (MNGIE type)	AR
UBA1	314370	VEXAS syndrome, somatic;Spinal muscular atrophy, XL 2, infantile	XLR
VAMP1	185880	Myasthenic syndrome, congenital, 25;Spastic ataxia 1, AD	AR, AD
VAPB	605704	Spinal muscular atrophy, late-onset, Finkel type;Amyotrophic lateral sclerosis 8	AD
VCP	601023	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1;Charcot-Marie-Tooth disease, type 2Y;Frontotemporal dementia and/or amyotrophic lateral sclerosis 6	AD

VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2	AR
VMA21	300913	Myopathy, XL, with excessive autophagy	XLR
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1	AR
VRK1	602168	Pontocerebellar hypoplasia type 1A	AR
WNK1	605232	Neuropathy, hereditary sensory and autonomic, type II;Pseudohypoaldosteronism, type IIC	AR, AD
XK	314850	McLeod syndrome with or without chronic granulomatous disease	XL
YARS1	603623	Charcot-Marie-Tooth disease, dominant intermediate C;Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2	AD, AR
ZC4H2	300897	Wieacker-Wolff syndrome;Wieacker-Wolff syndrome, female-restricted	XLR, XLD
DMPK	605377	Myotonic dystrophy 1	AD