(Neuro)Muscular panel

versie	03-12-2018 (215 genen)	Centrum voor Medische Genetica Gent

GitHub commit: e0a58ebf6cd19fa332a01b6b7109c050aee29f3e

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ABHD5	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ACADVL	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
ACTA1	102610	Myopathy, actin, congenital, with cores, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal recessive, Autosomal dominant; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal recessive, Autosomal dominant
ACVR1	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
AGL	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
AGRN	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive
ALDOA	103850	Glycogen storage disease XII, 611881 (3), Autosomal recessive
ALG13	300776	?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant
ALG14	612866	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3)
ALG2	607905	?Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
ALS2	606352	Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive; Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive
ANG	105850	Amyotrophic lateral sclerosis 9, 611895 (3)
ANO5	608662	Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2L, 611307 (3), Autosomal recessive

ASAH1	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
ATP2A1	108730	Brody myopathy, 601003 (3), Autosomal recessive
ATP7A	300011	Menkes disease, 309400 (3), X-linked recessive; Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
ATXN2	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Isolated cases, Multifactorial; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
BAG3	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
BICD2	609797	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 (3), Autosomal dominant
BIN1	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive
BSCL2	606158	Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant
BVES	604577	?Muscular dystrophy, limb-girdle, type 2X, 616812 (3), Autosomal recessive
C9orf72	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550 (3), Autosomal dominant
CAPN3	114240	Muscular dystrophy, limb-girdle, type 2A, 253600 (3), Autosomal recessive
CAV3	601253	Cardiomyopathy, familial hypertrophic, 192600 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type IC, 607801 (3), Autosomal recessive, Autosomal dominant; Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Rippling muscle disease, 606072 (3), Autosomal dominant
CAVIN1	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
CFL2	601443	Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive
CHAT	118490	Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive

CHKB	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHRNA1	100690	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal recessive, Autosomal dominant
CHRNB1	100710	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant; ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive
CHRND	100720	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive
CHRNE	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive
CHRNG	100730	Escobar syndrome, 265000 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
CLCN1	118425	Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia levior, recessive (3)
CNTN1	600016	?Myopathy, congenital, Compton-North, 612540 (3), Autosomal recessive
COL12A1	120320	Bethlem myopathy 2, 616471 (3), Autosomal dominant; ?Ullrich congenital muscular dystrophy 2, 616470 (3)
COL6A1	120220	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
COL6A2	120240	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; ?Myosclerosis, congenital, 255600 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
COL6A3	120250	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Dystonia 27, 616411 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
COLQ	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive

CPT2	600650	CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
CRYAB	123590	Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive
DAG1	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive
DAO	124050	No OMIM phenotype
DCTN1	601143	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Neuropathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant
DES	125660	Cardiomyopathy, dilated, 1I, 604765 (3); ?Muscular dystrophy, limb-girdle, type 2R, 615325 (3), Autosomal recessive; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant; Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant
DGUOK	601465	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive
DMD	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DMPK	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
DNAJB2	604139	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3), Autosomal recessive
DNAJB6	611332	Muscular dystrophy, limb-girdle, type 1E, 603511 (3), Autosomal dominant
DNM2	602378	Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant; Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive

DOK7	610285	?Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
DOLK	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
DPAGT1	191350	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
DPM1	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DPM2	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
DPM3	605951	Congenital disorder of glycosylation, type Io, 612937 (3)
DYNC1H1	600112	Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant; Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant
DYSF	603009	Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2B, 253601 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive
ECEL1	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
EMD	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
ENO3	131370	?Glycogen storage disease XIII, 612932 (3), Autosomal recessive
ETFA	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETFDH	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
FBLN5	604580	Cutis laxa, autosomal dominant 2, 614434 (3); Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant
FBXO38	608533	Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant
FDX2	614585	No OMIM phenotype

FHL1	300163	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive
FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive
FKTN	607440	Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive
FLNC	102565	Cardiomyopathy, familial hypertrophic, 26 (3); Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant
FUS	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
GAA	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
GARS	600287	Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant
GBE1	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
GFPT1	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
GLE1	603371	Arthrogryposis, lethal, with anterior horn cell disease, 611890 (3); Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive
GMPPB	615320	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive

GNE	603824	Nonaka myopathy, 605820 (3), Autosomal recessive; Sialuria, 269921 (3), Autosomal dominant
GRN	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
GYG1	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive
GYS1	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
HACD1	610467	No OMIM phenotype
HADHA	600890	Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive; HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Trifunctional protein deficiency, 609015 (3), Autosomal recessive
HADHB	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
HARS	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HNRNPA1	164017	Amyotrophic lateral sclerosis 20, 615426 (3), Autosomal dominant; ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 (3), Autosomal dominant
HNRNPA2B1	600124	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3)
HNRNPDL	607137	Muscular dystrophy, limb-girdle, type 1G, 609115 (3), Autosomal dominant
HSPB1	602195	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant
HSPB3	604624	?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant
HSPB8	608014	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive
ISCU	611911	Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive

ISPD	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive
ITGA7	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
KBTBD13	613727	Nemaline myopathy 6, autosomal dominant, 609273 (3), Autosomal dominant
KLHL40	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3)
KLHL41	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
KLHL9	611201	No OMIM phenotype
LAMA2	156225	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive
LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
LAMP2	309060	Danon disease, 300257 (3), X-linked dominant
LARGE1	603590	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive
LAS1L	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
LDB3	605906	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Left ventricular noncompaction 3, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant
LDHA	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
LIMS2	607908	Muscular dystrophy, limb-girdle, type 2W, 616827 (3), Autosomal recessive
LMNA	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, AD, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, AR, 616516 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 1B, 159001 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
LMOD3	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
LPIN1	605518	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive

LRP4	604270	Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant
MATR3	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
MEGF10	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 (3), Autosomal recessive; Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 (3), Autosomal recessive
MSTN	601788	Muscle hypertrophy, 614160 (3)
MTM1	300415	Myotubular myopathy, X-linked, 310400 (3), X-linked recessive
MUSK	601296	Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
MYBPC3	600958	Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
MYH14	608568	Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant; ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant
MYH3	160720	Arthrogryposis, distal, type 2A, 193700 (3), Autosomal dominant; Arthrogryposis, distal, type 2B, 601680 (3), Autosomal dominant; Arthrogryposis, distal, type 8, 178110 (3), Autosomal dominant
MYH7	160760	Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Autosomal dominant; Laing distal myopathy, 160500 (3), Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Scapuloperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant
MYH8	160741	Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant
MYOT	604103	Muscular dystrophy, limb-girdle, type 1A, 159000 (3), Autosomal dominant; Myopathy, myofibrillar, 3, 609200 (3), Autosomal dominant; Myopathy, spheroid body, 182920 (3), Autosomal dominant
NEB	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive
NEFH	162230	?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant
OPTN	602432	Amyotrophic lateral sclerosis 12, 613435 (3); Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3)

PABPN1	602279	Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant
PDE9A	602973	No OMIM phenotype
PFKM	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
PFN1	176610	Amyotrophic lateral sclerosis 18, 614808 (3)
PGAM2	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive
PGK1	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM1	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
PHKA1	311870	Muscle glycogenosis, 300559 (3), X-linked recessive
PHKB	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
PLEC	601282	Epidermolysis bullosa simplex with muscular dystrophy, 226670 (3), Autosomal recessive; ?Epidermolysis bullosa simplex with nail dystrophy, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2Q, 613723 (3), Autosomal recessive
PLEKHG5	611101	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive; Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive
PNPLA2	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
POLG	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 (3), Autosomal recessive
POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive; ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive

POMT1	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive
POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive
PREPL	609557	?Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
PRKAG2	602743	Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant; Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), ?Autosomal dominant
PYGM	608455	McArdle disease, 232600 (3), Autosomal recessive
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
RAPSN	601592	Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
REEP1	609139	?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant
RXYLT1 (TMEM5)	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
RYR1	180901	Central core disease, 117000 (3), Autosomal recessive, Autosomal dominant; King-Denborough syndrome, 145600 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 1}, 145600 (3), Autosomal dominant; Minicore myopathy with external ophthalmoplegia, 255320 (3), Autosomal recessive; Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 (3), Autosomal recessive, Autosomal dominant
SCN4A	603967	Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Paramyotonia congenita, 168300 (3), Autosomal dominant

SELENON	606210	Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant
SETX	608465	Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 1, 606002 (3), Autosomal recessive
SGCA	600119	Muscular dystrophy, limb-girdle, type 2D, 608099 (3), Autosomal recessive
SGCB	600900	Muscular dystrophy, limb-girdle, type 2E, 604286 (3), Autosomal recessive
SGCD	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, type 2F, 601287 (3), Autosomal recessive
SGCE	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
SGCG	608896	Muscular dystrophy, limb-girdle, type 2C, 253700 (3), Autosomal recessive
SIGMAR1	601978	?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive; ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive
SIL1	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SLC18A3	600336	Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive
SLC22A5	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
SLC52A2	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
SLC52A3	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
SLC5A7	608761	Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant
SMCHD1	614982	Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant; Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 (3)
SMN1	600354	Spinal muscular atrophy-1, 253300 (3), Autosomal recessive; Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive
SNAP25	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
SOD1	147450	Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant
SPEG	615950	Centronuclear myopathy 5, 615959 (3), Autosomal recessive

SQSTM1	601530	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Paget disease of bone 3, 167250 (3), Autosomal dominant
STAC3	615521	Native American myopathy, 255995 (3), Autosomal recessive
STIM1	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant
SYNE1	608441	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
SYNE2	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant
SYT2	600104	Myasthenic syndrome, congenital, 7, presynaptic, 616040 (3), Autosomal dominant
TAF15	601574	Chondrosarcoma, extraskeletal myxoid, 612237 (1)
TARDBP	605078	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant; Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant
TCAP	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2G, 601954 (3), Autosomal recessive
TFG	602498	Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant; ?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive
TIA1	603518	Welander distal myopathy, 604454 (3), Autosomal recessive, Autosomal dominant
TK2	188250	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 (3), Autosomal recessive; ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 (3), Autosomal recessive
TMEM43	612048	Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant
TNNI2	191043	Arthrogryposis multiplex congenita, distal, type 2B, 601680 (3), Autosomal dominant
TNNT1	191041	Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive
TNNT3	600692	Arthrogryposis, distal, type 2B, 601680 (3), Autosomal dominant
TNPO3	610032	Muscular dystrophy, limb-girdle, type 1F, 608423 (3), Autosomal dominant
TOR1AIP1	614512	?Muscular dystrophy, limb-girdle, type 2Y, 617072 (3), Autosomal recessive

TPM2	190990	Arthrogryposis multiplex congenita, distal, type 1, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 2B, 601680 (3), Autosomal dominant; CAP myopathy 2, 609285 (3), Autosomal dominant; Nemaline myopathy 4, autosomal dominant, 609285 (3), Autosomal dominant
TPM3	191030	CAP myopathy 1, 609284 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant; Nemaline myopathy 1, autosomal dominant or recessive, 609284 (3), Autosomal recessive, Autosomal dominant
TRAPPC11	614138	Muscular dystrophy, limb-girdle, type 2S, 615356 (3), Autosomal recessive
TRIM32	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2H, 254110 (3), Autosomal recessive
TRIM63	606131	No OMIM phenotype
TRPV4	605427	?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; SED, Maroteaux type, 184095 (3), Autosomal dominant; Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); Spinal muscular atrophy, distal, congenital nonprogressive, 600175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant
TSEN54	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
TTN	188840	Cardiomyopathy, dilated, 1G, 604145 (3); Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2J, 608807 (3), Autosomal recessive; Myopathy, proximal, with early respiratory muscle involvement, 603689 (3); Salih myopathy, 611705 (3), Autosomal recessive; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant
UBA1	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive
UBQLN2	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3), X-linked dominant
UNC13A	609894	No OMIM phenotype
VAPB	605704	Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant; Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant

VCP	601023	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3); Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant
VEGFA	192240	{Microvascular complications of diabetes 1}, 603933 (3)
VRK1	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: July 04, 2018

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.