

(Neuro)Muscular panel		
versie	v4 (312 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABHD5</i>	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
<i>ACADVL</i>	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
<i>ACTA1</i>	102610	Myopathy, actin, congenital, with cores, 161800 (3), Autosomal recessive, Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal recessive, Autosomal dominant; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant
<i>ACTN2</i>	102573	Myopathy, distal, 6, adult onset, 618655 (3), Autosomal dominant; Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant; Myopathy, congenital with structured cores and Z-line abnormalities, 618654 (3), Autosomal dominant
<i>ACVR1</i>	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
<i>ADCY6</i>	600294	?Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive
<i>ADGRG6</i>	612243	Lethal congenital contracture syndrome 9, 616503 (3), Autosomal recessive
<i>ADSS1</i> (<i>ADSSL1</i>)	612498	Myopathy, distal, 5, 617030 (3), Autosomal recessive
<i>AGL</i>	610860	Glycogen storage disease IIIb, 232400 (3), Autosomal recessive; Glycogen storage disease IIIa, 232400 (3), Autosomal recessive
<i>AGRN</i>	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive
<i>ALDOA</i>	103850	Glycogen storage disease XII, 611881 (3), Autosomal recessive
<i>ALG13</i>	300776	Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant; ?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked dominant
<i>ALG14</i>	612866	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive
<i>ALG2</i>	607905	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive; ?Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive

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<i>ALS2</i>	606352	Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive
<i>ANG</i>	105850	Amyotrophic lateral sclerosis 9, 611895 (3)
<i>ANOS</i>	608662	Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant
<i>ANXA11</i>	602572	Amyotrophic lateral sclerosis 23, 617839 (3), Autosomal dominant
<i>APOO</i>	300753	No OMIM phenotype
<i>ASAH1</i>	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
<i>ASCC1</i>	614215	Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
<i>ASPH</i>	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
<i>ATP2A1</i>	108730	Brody myopathy, 601003 (3), Autosomal recessive
<i>ATP7A</i>	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
<i>ATXN2</i>	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant
<i>B3GALNT2</i>	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
<i>B4GAT1</i>	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
<i>BAG3</i>	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
<i>BET1</i>	605456	No OMIM phenotype
<i>BICD2</i>	609797	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant
<i>BIN1</i>	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive
<i>BSCL2</i>	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive

H9.1-OP2-B9: Genpanel Neuromusculaire Dystrofie; in voege op 20/01/2021

<i>BVES</i>	604577	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 (3), Autosomal recessive
<i>C9orf72</i>	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550 (3), Autosomal dominant
<i>CACNA1H</i>	607904	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 (3); {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 (3); Hyperaldosteronism, familial, type IV, 617027 (3), Autosomal dominant
<i>CACNA1S</i>	114208	{Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant; {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; {Malignant hyperthermia, susceptibility to, 5}, 601887 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant
<i>CAPN3</i>	114240	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant
<i>CASQ1</i>	114250	Myopathy, vacuolar, with CASQ1 aggregates, 616231 (3), Autosomal dominant
<i>CAV3</i>	601253	Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant; Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 192600 (3), Digenic dominant, Autosomal dominant
<i>CAVIN1</i>	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
<i>CCDC78</i>	614666	?Centronuclear myopathy 4, 614807 (3), Autosomal dominant
<i>CFL2</i>	601443	Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive
<i>CHAT</i>	118490	Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive
<i>CHCHD10</i>	615903	Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant; ?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant
<i>CHKB</i>	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
<i>CHMP2B</i>	609512	Amyotrophic lateral sclerosis 17, 614696 (3), Autosomal dominant; Dementia, familial, nonspecific, 600795 (3), Autosomal dominant
<i>CHRNA1</i>	100690	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal recessive, Autosomal dominant; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive

<i>CHRNA1</i>	100710	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive; Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant
<i>CHRND</i>	100720	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
<i>CHRNE</i>	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive
<i>CHRNA3</i>	100730	Escobar syndrome, 265000 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
<i>CLCN1</i>	118425	Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia levior, recessive (3); Myotonia congenita, recessive, 255700 (3), Autosomal recessive
<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLPP</i>	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
<i>CNTN1</i>	600016	?Myopathy, congenital, Compton-North, 612540 (3), Autosomal recessive
<i>COL12A1</i>	120320	Bethlem myopathy 2, 616471 (3), Autosomal dominant; ?Ullrich congenital muscular dystrophy 2, 616470 (3)
<i>COL13A1</i>	120350	Myasthenic syndrome, congenital, 19, 616720 (3), Autosomal recessive
<i>COL4A1</i>	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant
<i>COL6A1</i>	120220	Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant; Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant
<i>COL6A2</i>	120240	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant; ?Myosclerosis, congenital, 255600 (3), Autosomal recessive
<i>COL6A3</i>	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
<i>COL9A3</i>	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; {Intervertebral disc disease, susceptibility to}, 603932 (3)
<i>COLQ</i>	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive
<i>CPT2</i>	600650	CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
<i>CRPPA (ISPD)</i>	614631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive
<i>CRYAB</i>	123590	Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant; Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant
<i>DAG1</i>	128239	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive
<i>DAO</i>	124050	No OMIM phenotype
<i>DCTN1</i>	601143	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant
<i>DES</i>	125660	?Cardiomyopathy, dilated, 1I, 604765 (3), Autosomal dominant; Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant
<i>DGUOK</i>	601465	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
<i>DHX16</i>	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant

<i>DMD</i>	300377	Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Becker muscular dystrophy, 300376 (3), X-linked recessive; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
<i>DMPK</i>	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
<i>DNAJB2</i>	604139	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3), Autosomal recessive
<i>DNAJB6</i>	611332	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511 (3), Autosomal dominant
<i>DNM2</i>	602378	Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant
<i>DNMT3B</i>	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
<i>DOK7</i>	610285	Fetal akinesia deformation sequence 3, 618389 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
<i>DOLK</i>	610746	Congenital disorder of glycosylation, type 1m, 610768 (3), Autosomal recessive
<i>DPAGT1</i>	191350	Congenital disorder of glycosylation, type 1j, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
<i>DPM1</i>	603503	Congenital disorder of glycosylation, type 1e, 608799 (3), Autosomal recessive
<i>DPM2</i>	603564	Congenital disorder of glycosylation, type 1u, 615042 (3), Autosomal recessive
<i>DPM3</i>	605951	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 (3), Autosomal recessive; ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 (3)
<i>DYNC1H1</i>	600112	Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant
<i>DYSF</i>	603009	Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive
<i>ECEL1</i>	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
<i>EMD</i>	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
<i>ENO3</i>	131370	?Glycogen storage disease XIII, 612932 (3), Autosomal recessive
<i>ERBB4</i>	600543	Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant

<i>ETFA</i>	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
<i>ETFB</i>	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
<i>ETFDH</i>	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
<i>EXOSC3</i>	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
<i>FAM111B</i>	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
<i>FBLN5</i>	604580	Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive
<i>FBXO38</i>	608533	Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant
<i>FDX2</i>	614585	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive
<i>FHL1</i>	300163	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Scapulooperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive
<i>FIG4</i>	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant
<i>FKBP14</i>	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive
<i>FKRP</i>	606596	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive
<i>FKTN</i>	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive

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<i>FLAD1</i>	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive
<i>FLNC</i>	102565	Cardiomyopathy, familial hypertrophic, 26, 617047 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant; Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant
<i>FUS</i>	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
<i>FXR1</i>	600819	?Myopathy, congenital proximal, with minicore lesions, 618823 (3), Autosomal recessive; ?Myopathy, congenital, with respiratory insufficiency and bone fractures, 618822 (3), Autosomal recessive
<i>GAA</i>	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
<i>GARS1 (GARS)</i>	600287	Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant
<i>GBE1</i>	607839	Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive; Glycogen storage disease IV, 232500 (3), Autosomal recessive
<i>GFER</i>	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3)
<i>GFPT1</i>	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
<i>GGPS1</i>	606982	No OMIM phenotype
<i>GLDN</i>	608603	Lethal congenital contracture syndrome 11, 617194 (3), Autosomal recessive
<i>GLE1</i>	603371	Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive; Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive
<i>GMPPB</i>	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
<i>GNE</i>	603824	Sialuria, 269921 (3), Autosomal dominant; Nonaka myopathy, 605820 (3), Autosomal recessive
<i>GOLGA2</i>	602580	No OMIM phenotype
<i>GOSR2</i>	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
<i>GRN</i>	138945	Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
<i>GYG1</i>	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive

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<i>GYS1</i>	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
<i>HACD1</i>	610467	No OMIM phenotype
<i>HADHA</i>	600890	LCHAD deficiency, 609016 (3), Autosomal recessive; HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
<i>HADHB</i>	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
<i>HARS1</i> (<i>HARS</i>)	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
<i>HEXB</i>	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
<i>HNRNPA1</i>	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
<i>HNRNPA2B1</i>	600124	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3)
<i>HNRNPD1</i>	607137	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115 (3), Autosomal dominant
<i>HRAS</i>	190020	Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant
<i>HSPB1</i>	602195	Neuronopathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant
<i>HSPB3</i>	604624	?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant
<i>HSPB8</i>	608014	Neuronopathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant
<i>HSPG2</i>	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
<i>HTRA2</i>	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
<i>IGHMBP2</i>	600502	Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive

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<i>INPP5K</i>	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
<i>ISCU</i>	611911	Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive
<i>ITGA7</i>	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
<i>KBTBD13</i>	613727	Nemaline myopathy 6, autosomal dominant, 609273 (3), Autosomal dominant
<i>KIF5A</i>	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant
<i>KLHL40</i>	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3), Autosomal recessive
<i>KLHL41</i>	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
<i>KLHL9</i>	611201	No OMIM phenotype
<i>KY</i>	605739	Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive
<i>LAMA2</i>	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
<i>LAMA5</i>	601033	No OMIM phenotype
<i>LAMB2</i>	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
<i>LAMP2</i>	309060	Danon disease, 300257 (3), X-linked dominant
<i>LARGE1</i>	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
<i>LARS2</i>	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
<i>LAS1L</i>	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
<i>LDB3</i>	605906	Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant; Left ventricular noncompaction 3, 601493 (3), Autosomal dominant
<i>LDHA</i>	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
<i>LIMS2</i>	607908	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 (3), Autosomal recessive

<i>LMNA</i>	150330	Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Malouf syndrome, 212112 (3), Autosomal dominant
<i>LMOD3</i>	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
<i>LOXL4</i>	607318	No OMIM phenotype
<i>LPIN1</i>	605518	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive
<i>LRIF1</i>	615354	No OMIM phenotype
<i>LRP4</i>	604270	?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive
<i>MAP3K20</i>	609479	Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive; Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive
<i>MATR3</i>	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
<i>MB</i>	160000	No OMIM phenotype
<i>MCOLN1</i>	605248	Mucopolipidosis IV, 252650 (3), Autosomal recessive
<i>MEGF10</i>	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 (3), Autosomal recessive; Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 (3), Autosomal recessive
<i>MET</i>	164860	{Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); ?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive; Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3)
<i>MICU1</i>	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
<i>MPDU1</i>	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
<i>MRPS25</i>	611987	No OMIM phenotype
<i>MSTN</i>	601788	Muscle hypertrophy, 614160 (3)
<i>MSTO1</i>	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal recessive, Autosomal dominant
<i>MTM1</i>	300415	Myotubular myopathy, X-linked, 310400 (3), X-linked recessive

<i>MTMR14</i>	611089	{Centronuclear myopathy, autosomal, modifier of}, 160150 (3), Autosomal dominant
<i>MUSK</i>	601296	Fetal akinesia deformation sequence 1, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
<i>MYBPC1</i>	160794	Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant; Myopathy, congenital, with tremor, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive
<i>MYBPC3</i>	600958	Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal recessive, Autosomal dominant; Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
<i>MYF5</i>	159990	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155 (3), Autosomal recessive
<i>MYH14</i>	608568	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant; Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant
<i>MYH2</i>	160740	Proximal myopathy and ophthalmoplegia, 605637 (3), Autosomal recessive, Autosomal dominant
<i>MYH3</i>	160720	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 (3), Autosomal recessive; Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 (3), Autosomal dominant; Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 (3), Autosomal dominant; Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant
<i>MYH7</i>	160760	Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Laing distal myopathy, 160500 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Scapuloperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant
<i>MYH8</i>	160741	Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant
<i>MYL1</i>	160780	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414 (3), Autosomal recessive
<i>MYMK</i>	615345	Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive
<i>MYO18B</i>	607295	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive
<i>MYO9A</i>	604875	Myasthenic syndrome, congenital, 24, presynaptic, 618198 (3), Autosomal recessive

<i>MYOT</i>	604103	Myopathy, myofibrillar, 3, 609200 (3), Autosomal dominant; Myopathy, spheroid body, 182920 (3), Autosomal dominant
<i>MYPN</i>	608517	Cardiomyopathy, dilated, 1KK, 615248 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 4, 615248 (3), Autosomal dominant; Nemaline myopathy 11, autosomal recessive, 617336 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 22, 615248 (3), Autosomal dominant
<i>NEB</i>	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive
<i>NEFH</i>	162230	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant; ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
<i>NEFL</i>	162280	Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
<i>NEK1</i>	604588	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant; Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive
<i>OPTN</i>	602432	{Glaucoma, normal tension, susceptibility to}, 606657 (3); Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; Amyotrophic lateral sclerosis 12, 613435 (3)
<i>ORAI1</i>	610277	Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant; Immunodeficiency 9, 612782 (3), Autosomal recessive
<i>PABPN1</i>	602279	Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant
<i>PAX7</i>	167410	Myopathy, congenital, progressive, with scoliosis, 618578 (3), Autosomal recessive; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
<i>PDE9A</i>	602973	No OMIM phenotype
<i>PEX6</i>	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive
<i>PFKM</i>	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
<i>PFN1</i>	176610	Amyotrophic lateral sclerosis 18, 614808 (3)
<i>PGAM2</i>	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive
<i>PGK1</i>	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
<i>PGM1</i>	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
<i>PHKA1</i>	311870	Muscle glycogenesis, 300559 (3), X-linked recessive
<i>PHKB</i>	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive

<i>PIEZO2</i>	613629	Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant; Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant
<i>PLEC</i>	601282	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive; Epidermolysis bullosa simplex with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex with muscular dystrophy, 226670 (3), Autosomal recessive; ?Epidermolysis bullosa simplex with nail dystrophy, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex, Ogna type, 131950 (3), Autosomal dominant
<i>PLEKHG5</i>	611101	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive
<i>PNPLA2</i>	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
<i>PNPLA8</i>	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
<i>POGLUT1</i>	615618	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive; Dowling-Degos disease 4, 615696 (3), Autosomal dominant
<i>POLG</i>	174763	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<i>POMGNT1</i>	606822	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; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive
<i>POMGNT2</i>	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive
<i>POMK</i>	615247	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive

<i>POMT1</i>	607423	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; 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
<i>POMT2</i>	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; 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
<i>POPDC3</i>	605824	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848 (3), Autosomal recessive
<i>PREPL</i>	609557	Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
<i>PRKAG2</i>	602743	Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), Autosomal dominant; Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant
<i>PRPH</i>	170710	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
<i>PTRH2</i>	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
<i>PUS1</i>	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
<i>PYGM</i>	608455	McArdle disease, 232600 (3), Autosomal recessive
<i>PYROXD1</i>	617220	Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive
<i>QDPR</i>	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
<i>RAPSN</i>	601592	Fetal akinesia deformation sequence 2, 618388 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive
<i>RBCK1</i>	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
<i>REEP1</i>	609139	Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant
<i>RXYLT1</i>	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive

<i>RYR1</i>	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
<i>RYR3</i>	180903	No OMIM phenotype
<i>SCN4A</i>	603967	Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant; Paramyotonia congenita, 168300 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant
<i>SELENON</i>	606210	Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant
<i>SEPTIN9 (SEPT9)</i>	604061	Amyotrophy, hereditary neuralgic, 162100 (3), Autosomal dominant
<i>SETX</i>	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
<i>SGCA</i>	600119	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 (3), Autosomal recessive
<i>SGCB</i>	600900	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 (3), Autosomal recessive
<i>SGCD</i>	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 (3), Autosomal recessive
<i>SGCE</i>	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
<i>SGCG</i>	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal recessive
<i>SIGMAR1</i>	601978	?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive; ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive
<i>SIL1</i>	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<i>SLC16A1</i>	600682	Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant
<i>SLC18A3</i>	600336	Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive
<i>SLC22A5</i>	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive

<i>SLC25A1</i>	190315	Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive; Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive
<i>SLC25A20</i>	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
<i>SLC25A4</i>	103220	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant
<i>SLC25A42</i>	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive
<i>SLC52A2</i>	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
<i>SLC52A3</i>	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
<i>SLC5A7</i>	608761	Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive
<i>SMCHD1</i>	614982	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 (3); Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant
<i>SMN1</i>	600354	Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-1, 253300 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive
<i>SMPX</i>	300226	Deafness, X-linked 4, 300066 (3), X-linked dominant
<i>SNAP25</i>	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
<i>SOD1</i>	147450	Spastic tetraplegia and axial hypotonia, progressive, 618598 (3), Autosomal recessive; Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant
<i>SPEG</i>	615950	Centronuclear myopathy 5, 615959 (3), Autosomal recessive
<i>SPG11</i>	610844	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive; Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive
<i>SPTBN4</i>	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive

<i>SQSTM1</i>	601530	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
<i>STAC3</i>	615521	Myopathy, congenital, Baily-Bloch, 255995 (3), Autosomal recessive
<i>STIM1</i>	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive; Stormorken syndrome, 185070 (3), Autosomal dominant
<i>SVIL</i>	604126	No OMIM phenotype
<i>SYNE1</i>	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant
<i>SYNE2</i>	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant
<i>SYT2</i>	600104	Myasthenic syndrome, congenital, 7, presynaptic, 616040 (3), Autosomal dominant
<i>TAF15</i>	601574	Chondrosarcoma, extraskeletal myxoid, 612237 (1)
<i>TARDBP</i>	605078	Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant; Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant
<i>TBK1</i>	604834	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant; {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant
<i>TCAP</i>	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive
<i>TFAM</i>	600438	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 (3), Autosomal recessive
<i>TFG</i>	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
<i>TIA1</i>	603518	Welander distal myopathy, 604454 (3), Autosomal recessive, Autosomal dominant
<i>TIMM22</i>	607251	?Combined oxidative phosphorylation deficiency 43, 618851 (3), Autosomal recessive
<i>TK2</i>	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

H9.1-OP2-B9: Genpanel Neuromusculaire Dystrofie; in voege op 20/01/2021

<i>TMEM43</i>	612048	Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant
<i>TMEM65</i>	616609	No OMIM phenotype
<i>TNNC2</i>	191039	No OMIM phenotype
<i>TNNI2</i>	191043	Arthrogryposis, distal, type 2B1, 601680 (3), Autosomal dominant
<i>TNNT1</i>	191041	Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive
<i>TNNT3</i>	600692	Arthrogryposis, distal, type 2B2, 618435 (3), Autosomal dominant
<i>TNPO3</i>	610032	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423 (3), Autosomal dominant
<i>TNXB</i>	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
<i>TOR1AIP1</i>	614512	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 (3), Autosomal recessive
<i>TPM2</i>	190990	Nemaline myopathy 4, autosomal dominant, 609285 (3), Autosomal dominant; Arthrogryposis, distal, type 2B4, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 1A, 108120 (3), Autosomal dominant; CAP myopathy 2, 609285 (3), Autosomal dominant
<i>TPM3</i>	191030	CAP myopathy 1, 609284 (3), Autosomal recessive, Autosomal dominant; Nemaline myopathy 1, autosomal dominant or recessive, 609284 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant
<i>TRAPPC11</i>	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
<i>TRIM54</i>	606474	No OMIM phenotype
<i>TRIM63</i>	606131	No OMIM phenotype
<i>TRIP4</i>	604501	Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive; ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive

<i>TRPV4</i>	605427	Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; SED, Maroteaux type, 184095 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIII, 600175 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant
<i>TSEN54</i>	608755	Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
<i>TTN</i>	188840	Myopathy, myofibrillar, 9, with early respiratory failure, 603689 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 (3), Autosomal recessive; Cardiomyopathy, dilated, 1G, 604145 (3); Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant; Salih myopathy, 611705 (3), Autosomal recessive
<i>TUBA4A</i>	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant
<i>UBA1</i>	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive
<i>UBQLN2</i>	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3), X-linked dominant
<i>UNC13A</i>	609894	No OMIM phenotype
<i>UNC45B</i>	611220	?Cataract 43, 616279 (3), Autosomal dominant
<i>VAMP1</i>	185880	Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant; Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive
<i>VAPB</i>	605704	Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant
<i>VCP</i>	601023	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3)
<i>VEGFA</i>	192240	{Microvascular complications of diabetes 1}, 603933 (3)
<i>VMA21</i>	300913	Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive
<i>VPS33B</i>	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive

<i>VRK1</i>	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
<i>YARS2</i>	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive
<i>ZBTB42</i>	613915	?Lethal congenital contracture syndrome 6, 616248 (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: Sep 01, 2020

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.