

<b>NeuroMuscular panel</b>		
<b>versie</b>	26-Feb-2018 (216 genen)	Centrum voor Medische Genetica Gent
<b>Gene</b>	<b>OMIM gene ID</b>	<b>Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern</b>
<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; 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
<i>ACVR1</i>	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
<i>AGL</i>	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 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	?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant
<i>ALG14</i>	612866	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3)
<i>ALG2</i>	607905	?Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
<i>ALS2</i>	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
<i>ANG</i>	105850	Amyotrophic lateral sclerosis 9, 611895 (3)
<i>ANO5</i>	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
<i>ASAH1</i>	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive

<i>ATP2A1</i>	108730	Brody myopathy, 601003 (3), Autosomal recessive
<i>ATP7A</i>	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
<i>ATXN2</i>	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
<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>BICD2</i>	609797	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 (3), Autosomal dominant
<i>BIN1</i>	601248	Myopathy, centronuclear, autosomal recessive, 255200 (3), Autosomal recessive
<i>BSCL2</i>	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
<i>BVES</i>	604577	?Muscular dystrophy, limb-girdle, type 2X, 616812 (3), Autosomal recessive
<i>C9orf72</i>	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550 (3), Autosomal dominant
<i>CAPN3</i>	114240	Muscular dystrophy, limb-girdle, type 2A, 253600 (3), Autosomal recessive
<i>CAV3</i>	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
<i>CAVIN1</i>	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
<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>CHKB</i>	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
<i>CHRNA1</i>	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
<i>CHRNB1</i>	100710	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant; ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive
<i>CHRND</i>	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
<i>CHRNE</i>	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
<i>CHRNA1</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 congenita, recessive, 255700 (3), Autosomal recessive; Myotonia levior, recessive (3)
<i>CNTN1</i>	600016	?Myopathy, congenital, Compton-North, 612540 (3), Autosomal recessive
<i>COL12A1</i>	120320	Bethlem myopathy 2, 616471 (3); ?Ullrich congenital muscular dystrophy 2, 616470 (3)
<i>COL6A1</i>	120220	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
<i>COL6A2</i>	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
<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>COLQ</i>	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive

<i>CPT2</i>	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
<i>CRYAB</i>	123590	Cardiomyopathy, dilated, 11I, 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
<i>DAG1</i>	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
<i>DAO</i>	124050	{Schizophrenia}, 181500 (2), Autosomal dominant
<i>DCTN1</i>	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
<i>DES</i>	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; Scapulooperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant
<i>DGUOK</i>	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
<i>DMD</i>	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; 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, type 1E, 603511 (3), Autosomal dominant
<i>DNM2</i>	602378	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; Myopathy, centronuclear 1, 160150 (3), Autosomal dominant

<i>DOK7</i>	610285	?Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
<i>DOLK</i>	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
<i>DPAGT1</i>	191350	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
<i>DPM1</i>	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
<i>DPM2</i>	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
<i>DPM3</i>	605951	Congenital disorder of glycosylation, type Io, 612937 (3)
<i>DYNC1H1</i>	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
<i>DYSF</i>	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
<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>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>FBLN5</i>	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
<i>FBXO38</i>	608533	Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant
<i>FDX1L</i> ( <i>FDX2</i> )	614585	No OMIM phenotype

<i>FHL1</i>	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; Scapulooperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant
<i>FKRP</i>	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
<i>FKTN</i>	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
<i>FLNC</i>	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
<i>FUS</i>	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Tremor, hereditary essential, 4, 614782 (3), Autosomal dominant
<i>GAA</i>	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
<i>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	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
<i>GFPT1</i>	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
<i>GLE1</i>	603371	Arthrogryposis, lethal, with anterior horn cell disease, 611890 (3); 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	Nonaka myopathy, 605820 (3), Autosomal recessive; Sialuria, 269921 (3), Autosomal dominant
<i>GRN</i>	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
<i>GYG1</i>	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive
<i>GYS1</i>	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
<i>HACD1</i>	610467	No OMIM phenotype
<i>HADHA</i>	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
<i>HADHB</i>	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
<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, type 1G, 609115 (3), Autosomal dominant
<i>HSPB1</i>	602195	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant
<i>HSPB3</i>	604624	?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant
<i>HSPB8</i>	608014	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIA, 158590 (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>IGHMBP2</i>	600502	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive
<i>ISCU</i>	611911	Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive
<i>ISPD</i>	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
<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>KLHL40</i>	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3)
<i>KLHL41</i>	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
<i>KLHL9</i>	611201	No OMIM phenotype
<i>LAMA2</i>	156225	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive
<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 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
<i>LAS1L</i>	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
<i>LDB3</i>	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
<i>LDHA</i>	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
<i>LIMS2</i>	607908	Muscular dystrophy, limb-girdle, type 2W, 616827 (3), Autosomal recessive



<i>LMNA</i>	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
<i>LMOD3</i>	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
<i>LPIN1</i>	605518	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive
<i>LRP4</i>	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
<i>MATR3</i>	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
<i>MEGF10</i>	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
<i>MSTN</i>	601788	Muscle hypertrophy, 614160 (3)
<i>MTM1</i>	300415	Myotubular myopathy, X-linked, 310400 (3), X-linked recessive
<i>MUSK</i>	601296	Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
<i>MYBPC3</i>	600958	Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
<i>MYH14</i>	608568	Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant; ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant
<i>MYH3</i>	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

<i>MYH7</i>	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
<i>MYH8</i>	160741	Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant
<i>MYOT</i>	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
<i>NEB</i>	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive
<i>NEFH</i>	162230	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant
<i>OBSCN</i>	No OMIM gene	No OMIM phenotype
<i>OPTN</i>	602432	Amyotrophic lateral sclerosis 12, 613435 (3); Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3)
<i>PABPN1</i>	602279	Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant
<i>PDE9A</i>	602973	No OMIM phenotype
<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 glycogenosis, 300559 (3), X-linked recessive
<i>PHKB</i>	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
<i>PLEC</i>	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, Ogn type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2Q, 613723 (3), Autosomal recessive
<i>PLEKHG5</i>	611101	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive; Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive

<i>PNPLA2</i>	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
<i>POLG</i>	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
<i>POMGNT1</i>	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
<i>POMGNT2</i>	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive
<i>POMK</i>	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
<i>POMT1</i>	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
<i>POMT2</i>	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
<i>PREPL</i>	609557	?Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
<i>PRKAG2</i>	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
<i>PYGM</i>	608455	McArdle disease, 232600 (3), Autosomal recessive
<i>QDPR</i>	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive

<i>RAPSN</i>	601592	Fetal akinesia deformation sequence, 208150 (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	?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant
<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>SCN4A</i>	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
<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>SETX</i>	608465	Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 1, 606002 (3), Autosomal recessive
<i>SGCA</i>	600119	Muscular dystrophy, limb-girdle, type 2D, 608099 (3), Autosomal recessive
<i>SGCB</i>	600900	Muscular dystrophy, limb-girdle, type 2E, 604286 (3), Autosomal recessive
<i>SGCD</i>	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, type 2F, 601287 (3), Autosomal recessive
<i>SGCE</i>	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
<i>SGCG</i>	608896	Muscular dystrophy, limb-girdle, type 2C, 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>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>SLC25A20</i>	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (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	Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant
<i>SMCHD1</i>	614982	Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant; Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 (3)
<i>SMN1</i>	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
<i>SNAP25</i>	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
<i>SOD1</i>	147450	Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant
<i>SPEG</i>	615950	Centronuclear myopathy 5, 615959 (3), Autosomal recessive
<i>SQSTM1</i>	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
<i>STAC3</i>	615521	Native American myopathy, 255995 (3), Autosomal recessive
<i>STIM1</i>	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant
<i>SYNE1</i>	608441	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
<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	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant; Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant

<i>TCAP</i>	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2G, 601954 (3), Autosomal recessive
<i>TFG</i>	602498	Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant; ?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive
<i>TIA1</i>	603518	Welander distal myopathy, 604454 (3), Autosomal recessive, Autosomal dominant
<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
<i>TMEM43</i>	612048	Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant
<i>TMEM5</i>	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
<i>TNNI2</i>	191043	Arthrogryposis multiplex congenita, distal, type 2B, 601680 (3), Autosomal dominant
<i>TNNT1</i>	191041	Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive
<i>TNNT3</i>	600692	Arthrogryposis, distal, type 2B, 601680 (3), Autosomal dominant
<i>TNPO3</i>	610032	Muscular dystrophy, limb-girdle, type 1F, 608423 (3), Autosomal dominant
<i>TOR1AIP1</i>	614512	?Muscular dystrophy, limb-girdle, type 2Y, 617072 (3), Autosomal recessive
<i>TPM2</i>	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
<i>TPM3</i>	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
<i>TRAPPC11</i>	614138	Muscular dystrophy, limb-girdle, type 2S, 615356 (3), Autosomal recessive
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2H, 254110 (3), Autosomal recessive
<i>TRIM63</i>	606131	No OMIM phenotype

<i>TRPV4</i>	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; Scapulo-peroneal 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
<i>TSEN54</i>	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
<i>TTN</i>	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
<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>VAPB</i>	605704	Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant; Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant
<i>VCP</i>	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
<i>VEGFA</i>	192240	{Microvascular complications of diabetes 1}, 603933 (3)
<i>VRK1</i>	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: June 06, 2017

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.