

| NeuroMuscular panel | | |
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| versie | v5 (272 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>ACAD9</i> | 611103 | Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive |
| <i>ACADL</i> | 609576 | No OMIM phenotype |
| <i>ACADM</i> | 607008 | Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive |
| <i>ACADS</i> | 606885 | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (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 (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 |

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| <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 |
| <i>AMPD1</i> | 102770 | Myopathy due to myoadenylate deaminase deficiency, 615511 (3), Autosomal recessive |
| <i>ANO5</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>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>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>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 |
| <i>BVES</i> | 604577 | Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 (3), Autosomal recessive |
| <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 |

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| <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>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 |

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| <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 |

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| <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>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>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 |

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| <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 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 | 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>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>EPG5</i> | 615068 | Vici syndrome, 242840 (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>FAM111B</i> | 615584 | Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (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; Scapulo-peroneal 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>FKBP14</i> | 614505 | Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive |

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| <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 |
| <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>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>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 |

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| <i>GOSR2</i> | 604027 | Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive |
| <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 | 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>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>HNRNPDL</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 |

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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>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>LGI4</i> | 608303 | Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive |
| <i>LIMS2</i> | 607908 | ?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 (3), Autosomal recessive |

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| <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>MB</i> | 160000 | No OMIM phenotype |
| <i>MCOLN1</i> | 605248 | Mucopolidosis 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 |

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| <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; Scapulooperoneal 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>MYL2</i> | 160781 | Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant |
| <i>MYMK</i> | 615345 | Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive |

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| <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>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>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>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>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>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 |

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| <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>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>POLG2</i> | 604983 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (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 |

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| <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>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>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>RRM2B</i> | 604712 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive |
| <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 |

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| <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>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>SGCG</i> | 608896 | Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (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>SLC5A7</i> | 608761 | Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive |

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| <i>SMCHD1</i> | 614982 | Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 (3); Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant |
| <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>SPEG</i> | 615950 | Centronuclear myopathy 5, 615959 (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>SUCLA2</i> | 603921 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive |
| <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>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>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>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 |

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| <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 | 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>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>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 |

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| <i>TSMF</i> | 604723 | Combined oxidative phosphorylation deficiency 3, 610505 (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>TYMP</i> | 131222 | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive |
| <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>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>VMA21</i> | 300913 | Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive |
| <i>VPS33B</i> | 608552 | Arthrogyriposis, renal dysfunction, and cholestasis 1, 208085 (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 |
| <i>ZC4H2</i> | 300897 | Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant |

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.