NeuroMuscular

Gene panel

Gene panel information

Gene panel	**NeuroMuscular**
Version	7
Total genes	378
Activation date	Tuesday 15 october 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AARS1	99.99 %	601065	Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive
ABCC9	99.92 %	601439	Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive
ABHD5	99.98 %	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ACAD9	100 %	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
ACADL	99.11 %	609576	*No OMIM phenotypes*
ACADM	96.14 %	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
ACADS	99.99 %	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
ACADVL	100 %	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
ACTA1	99.99 %	102610	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265 (3), Autosomal recessive; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Congenital myopathy 2C, severe infantile, autosomal dominant, 620278 (3), Autosomal dominant; Congenital myopathy 2A, typical, autosomal dominant, 161800 (3), Autosomal dominant
ACTN2	99.99 %	102573	Myopathy, distal, 6, adult onset, 618655 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant; Congenital myopathy 8, 618654 (3), Autosomal dominant; Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant
ACVR1	99.94 %	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ADAMTS15	99.98 %	607509	Arthrogryposis, distal, type 12, 620545 (3), Autosomal recessive
ADCY6	99.98 %	600294	Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive
ADGRG6	99.91 %	612243	Lethal congenital contracture syndrome 9, 616503 (3), Autosomal recessive
ADSS1	99.99 %	612498	Myopathy, distal, 5, 617030 (3), Autosomal recessive
AGL	97.67 %	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
AGRN	99.99 %	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive
AK3	99.92 %	609290	*No OMIM phenotypes*
ALDOA	100 %	103850	Glycogen storage disease XII, 611881 (3), Autosomal recessive

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ALG13	99.44 %	300776	Developmental and epileptic encephalopathy 36, 300884 (3), X-linked
ALG14	99.34 %	612866	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 (3), Autosomal recessive; Myopathy, epilepsy, and progressive cerebral atrophy, 619036 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive
ALG2	100 %	607905	Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
AMPD1	98.94 %	102770	Myopathy due to myoadenylate deaminase deficiency, 615511 (3), Autosomal recessive
ANO5	99.85 %	608662	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant
ANXA11	99.71 %	602572	Amyotrophic lateral sclerosis 23, 617839 (3), Autosomal dominant; Inclusion body myopathy and brain white matter abnormalities, 619733 (3), Autosomal dominant
APOO	99.69 %	300753	*No OMIM phenotypes*
ASAH1	99.9 %	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
ASCC1	90.99 %	614215	Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
ASCC3	99.76 %	614217	Intellectual developmental disorder, autosomal recessive 81, 620700 (3), Autosomal recessive
ASPH	99.92 %	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
ATP1A2	99.85 %	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP2A1	99.83 %	108730	Brody myopathy, 601003 (3), Autosomal recessive
ATP2A2	99.98 %	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP7A	99.87 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
B3GALNT2	92.79 %	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive
B4GALNT1	99.97 %	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
B4GAT1	100 %	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
BAG3	100 %	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
BET1	99.38 %	605456	Muscular dystrophy, congenital, with rapid progression, 254100 (3), Autosomal recessive
BICD2	99.99 %	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
BIN1	99.98 %	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive

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BSCL2	99.99 %	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
BVES	99.83 %	604577	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 (3), Autosomal recessive
CACNA1H	100 %	607904	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 (3); Hyperaldosteronism, familial, type IV, 617027 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 (3)
CACNA1S	99.96 %	114208	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; Congenital myopathy 18 due to dihydropyridine receptor defect, 620246 (3), Autosomal dominant, Autosomal recessive; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant
CAP2	99.99 %	618385	Cardiomyopathy, dilated, 2I, 620462 (3), Autosomal recessive
CAPN1	99.99 %	114220	Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive
CAPN3	99.99 %	114240	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant
CASQ1	99.62 %	114250	Myopathy, vacuolar, with CASQ1 aggregates, 616231 (3), Autosomal dominant
CAV3	100 %	601253	Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 192600 (3), Digenic dominant, Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant
CAVIN1	100 %	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
CCDC78	100 %	614666	?Centronuclear myopathy 4, 614807 (3), Autosomal dominant
CFL2	99.67 %	601443	Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive
CHAT	99.74 %	118490	Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive
CHCHD10	100 %	615903	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant; Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant
CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHD8	99.96 %	610528	Intellectual developmental disorder with autism and macrocephaly, 615032 (3), Autosomal dominant
CHKB	100 %	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHRNA1	99.82 %	100690	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal dominant, Autosomal recessive; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
CHRNB1	99.99 %	100710	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive; Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CHRND	100 %	100720	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant
CHRNE	100 %	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal dominant, Autosomal recessive; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive
CHRNG	100 %	100730	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Escobar syndrome, 265000 (3), Autosomal recessive
CIAO1	99.88 %	604333	*No OMIM phenotypes*
CLCN1	100 %	118425	Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia levior, 160800 (3), Autosomal dominant
CLN3	99.92 %	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLPP	99.99 %	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
CNTN1	99.35 %	600016	Congenital myopathy 12, 612540 (3), Autosomal recessive
COA8	99.94 %	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
COASY	99.98 %	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COL12A1	99.85 %	120320	Bethlem myopathy 2, 616471 (3), Autosomal dominant; ?Ullrich congenital muscular dystrophy 2, 616470 (3), Autosomal recessive
COL13A1	99.95 %	120350	Myasthenic syndrome, congenital, 19, 616720 (3), Autosomal recessive
COL25A1	99.87 %	610004	Fibrosis of extraocular muscles, congenital, 5, 616219 (3), Autosomal recessive
COL4A1	99.99 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
COL6A1	99.99 %	120220	Ullrich congenital muscular dystrophy 1A, 254090 (3), Autosomal dominant, Autosomal recessive; Bethlem myopathy 1A, 158810 (3), Autosomal dominant
COL6A2	100 %	120240	?Myosclerosis, congenital, 255600 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1B, 620727 (3), Autosomal dominant, Autosomal recessive; Bethlem myopathy 1B, 620725 (3), Autosomal dominant, Autosomal recessive
COL6A3	99.99 %	120250	Bethlem myopathy 1C, 620726 (3), Autosomal dominant, Autosomal recessive; Ullrich congenital muscular dystrophy 1C, 620728 (3), Autosomal dominant, Autosomal recessive; Dystonia 27, 616411 (3), Autosomal recessive
COL9A3	99.99 %	120270	{Intervertebral disc disease, susceptibility to}, 603932 (3); Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; Stickler syndrome, type VI, 620022 (3), Autosomal recessive
COLQ	99.98 %	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive
COQ4	100 %	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive; Spastic ataxia 10, autosomal recessive, 620666 (3), Autosomal recessive
COQ7	100 %	601683	Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
COQ8A	100 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COX16	100 %	618064	Mitochondrial complex IV deficiency, nuclear type 22, 619355 (3), Autosomal recessive
COX6A2	99.75 %	602009	Mitochondrial complex IV deficiency, nuclear type 18, 619062 (3), Autosomal recessive
CPT2	99.65 %	600650	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal dominant, Autosomal recessive; 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 dominant, Autosomal recessive
CRPPA	99.98 %	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
CRYAB	100 %	123590	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant
CTBP1	99.98 %	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
DAG1	100 %	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
DCTN1	99.98 %	601143	Perry syndrome, 168605 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal dominant, Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641 (3), Autosomal dominant
DES	100 %	125660	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Cardiomyopathy, dilated, 1I, 604765 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal dominant, Autosomal recessive
DGUOK	99.93 %	601465	Portal hypertension, noncirrhotic, 1, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
DHX16	99.98 %	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant
DMD	99.76 %	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DNAJB2	99.97 %	604139	Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881 (3), Autosomal recessive
DNAJB4	99.06 %	611327	Congenital myopathy 21 with early respiratory failure, 620326 (3), Autosomal recessive
DNAJB6	99.97 %	611332	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511 (3), Autosomal dominant
DNM2	99.99 %	602378	Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant; Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DNMT3B	99.98 %	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant
DOK7	99.97 %	610285	Fetal akinesia deformation sequence 3, 618389 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
DOLK	100 %	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
DPAGT1	100 %	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive
DPM1	90.68 %	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DPM2	100 %	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
DPM3	99.98 %	605951	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 (3), Autosomal recessive
DTNA	100 %	601239	Left ventricular noncompaction 1, with or without congenital heart defects, 604169 (3), Autosomal dominant
DYNC1H1	99.99 %	600112	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 13, 614563 (3), Autosomal dominant
DYSF	99.95 %	603009	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive
EBF3	99.99 %	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
ECEL1	100 %	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
EMD	99.93 %	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
EMILIN1	99.99 %	130660	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080 (3), Autosomal dominant; Arterial tortuosity-bone fragility syndrome, 620908 (3), Autosomal recessive
ENDOG	99.99 %	600440	*No OMIM phenotypes*
ENO3	100 %	131370	Glycogen storage disease XIII, 612932 (3), Autosomal recessive
EPG5	99.95 %	615068	Vici syndrome, 242840 (3), Autosomal recessive
ETFA	99.88 %	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	100 %	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETFDH	99.82 %	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
EXOSC3	100 %	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EXOSC8	99.91 %	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
EXOSC9	94.91 %	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
FAM111B	99.98 %	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FBXO38	99.96 %	608533	Neuronopathy, distal hereditary motor, autosomal dominant 6, 615575 (3), Autosomal dominant
FDX2	99.99 %	614585	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive

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FHL1	99.97 %	300163	Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; Scapuloperoneal 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; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant
FILIP1	99.98 %	607307	Neuromuscular disorder, congenital, with dysmorphic facies, 620775 (3), Autosomal recessive
FKBP14	99.97 %	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive
FKRP	100 %	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive
FKTN	99.94 %	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 (3), Autosomal recessive
FLAD1	99.98 %	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive
FLNC	99.99 %	102565	Cardiomyopathy, familial hypertrophic, 26, 617047 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia, familial, 617047 (3), Autosomal dominant; Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant
FXR1	99.82 %	600819	Congenital myopathy 9B, proximal, with minicore lesions, 618823 (3), Autosomal recessive; ?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822 (3), Autosomal recessive
GAA	100 %	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
GARS1	99.93 %	600287	Spinal muscular atrophy, infantile, James type, 619042 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant
GBE1	99.73 %	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
GBF1	99.97 %	603698	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483 (3), Autosomal dominant
GFER	100 %	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive
GFPT1	99.79 %	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
GGPS1	99.95 %	606982	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518 (3), Autosomal recessive
GLDN	99.98 %	608603	Lethal congenital contracture syndrome 11, 617194 (3), Autosomal recessive
GLE1	99.99 %	603371	Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive; Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GMPPB	100 %	615320	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive
GNE	99.99 %	603824	Sialuria, 269921 (3), Autosomal dominant; Thrombocytopenia 12 with or without myopathy, 620757 (3), Autosomal recessive; Nonaka myopathy, 605820 (3), Autosomal recessive
GOLGA2	100 %	602580	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240 (3), Autosomal recessive
GOSR2	98.92 %	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive; Muscular dystrophy, congenital, with or without seizures, 620166 (3), Autosomal recessive
GRIN1	100 %	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
GYG1	99.85 %	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive
GYS1	99.98 %	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
HACD1	99.98 %	610467	Congenital myopathy 11, 619967 (3), Autosomal recessive
HADH	99.86 %	601609	Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive
HADHA	99.98 %	600890	HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency 1, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
HADHB	99.82 %	143450	Mitochondrial trifunctional protein deficiency 2, 620300 (3)
HEXB	99.91 %	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HINT1	99.86 %	601314	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive
HMGCR	99.73 %	142910	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375 (3), Autosomal recessive; [Statins, response to], 620410 (3); [Low density lipoprotein cholesterol level QTL 3], 620410 (3)
HMGCS1	99.98 %	142940	*No OMIM phenotypes*
HNRNPA1	62.92 %	164017	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 (3), Autosomal dominant; ?Myopathy, distal, 3, 610099 (3), Autosomal dominant; Amyotrophic lateral sclerosis 20, 615426 (3), Autosomal dominant
HNRNPA2B1	99.9 %	600124	Oculopharyngeal muscular dystrophy 2, 620460 (3), Autosomal dominant; ?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3), Autosomal dominant
HNRNPDL	99.98 %	607137	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115 (3), Autosomal dominant
HNRNPK	99.93 %	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HRAS	100 %	190020	Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant
HSPB1	99.97 %	602195	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634 (3), Autosomal dominant
HSPB3	99.97 %	604624	?Neuronopathy, distal hereditary motor, autosomal dominant 4, 613376 (3), Autosomal dominant
HSPB6	100 %	610695	*No OMIM phenotypes*
HSPB8	100 %	608014	Neuronopathy, distal hereditary motor, autosomal dominant 2, 158590 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant
HSPG2	99.87 %	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
HTRA2	99.99 %	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
IGHMBP2	99.92 %	600502	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320 (3), Autosomal recessive
INPP5K	99.94 %	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
ISCU	99.74 %	611911	Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive
ITGA7	99.87 %	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
JAG2	99.99 %	602570	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566 (3), Autosomal recessive
JPH1	99.98 %	605266	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, modifier of, 607831 (3), Autosomal dominant, Autosomal recessive
KBTBD13	100 %	613727	Nemaline myopathy 6, autosomal dominant, 609273 (3), Autosomal dominant
KCNJ2	100 %	600681	Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Andersen syndrome, 170390 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3), Autosomal dominant
KIF21A	96.41 %	608283	Fibrosis of extraocular muscles, congenital, 3B, 135700 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 1, 135700 (3), Autosomal dominant
KLHL40	99.96 %	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3), Autosomal recessive
KLHL41	99.96 %	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
KLHL9	100 %	611201	*No OMIM phenotypes*
KY	99.97 %	605739	Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive
LAMA2	99.95 %	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
LAMA5	99.99 %	601033	Nephrotic syndrome, type 26, 620049 (3), Autosomal recessive; ?Bent bone dysplasia syndrome 2, 620076 (3), Autosomal recessive
LAMB2	99.99 %	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3), Autosomal recessive; Pierson syndrome, 609049 (3), Autosomal recessive
LAMP2	98.95 %	309060	Danon disease, 300257 (3), X-linked dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LARGE1	100 %	603590	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
LARS2	99.96 %	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
LAS1L	99.97 %	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
LDB3	99.91 %	605906	Left ventricular noncompaction 3, 601493 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant; Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant
LDHA	99.94 %	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
LETM1	99.97 %	604407	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 (3), Autosomal recessive
LGI4	99.98 %	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive
LIG3	99.99 %	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
LIMS2	99.97 %	607908	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 (3), Autosomal recessive
LMNA	99.96 %	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
LMOD3	99.91 %	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
LOXL4	99.92 %	607318	*No OMIM phenotypes*
LPIN1	99.96 %	605518	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive
LRIF1	99.82 %	615354	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477 (3), Digenic recessive
LRP4	99.89 %	604270	?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal dominant, Autosomal recessive; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive
MAMDC2	100 %	612879	*No OMIM phenotypes*
MAP3K20	99.84 %	609479	Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive
MATR3	99.86 %	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
MB	99.99 %	160000	Myopathy, sarcoplasmic body, 620286 (3), Autosomal dominant
MCOLN1	100 %	605248	Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucolipidosis IV, 252650 (3), Autosomal recessive
MEGF10	99.91 %	612453	Congenital myopathy 10A, severe variant, 614399 (3), Autosomal recessive; Congenital myopathy 10B, mild variant, 620249 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MET	99.97 %	164860	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3); ?Arthrogryposis, distal, type 11, 620019 (3), Autosomal dominant; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); {Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; ?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive
MICU1	99.56 %	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MLIP	99.71 %	614106	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138 (3), Autosomal recessive
MPDU1	99.97 %	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MRPS25	99.97 %	611987	?Combined oxidative phosphorylation deficiency 50, 619025 (3), Autosomal recessive
MSTN	99.96 %	601788	?Muscle hypertrophy, 614160 (3), Autosomal recessive
MSTO1	76.34 %	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive
MTM1	99.76 %	300415	Myopathy, centronuclear, X-linked, 310400 (3), X-linked recessive
MTMR14	99.99 %	611089	{Centronuclear myopathy, autosomal, modifier of}, 160150 (3), Autosomal dominant
MUSK	99.93 %	601296	Fetal akinesia deformation sequence 1, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
MYBPC1	99.77 %	160794	Congenital myopathy 16, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive; Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant
MYBPC3	99.98 %	600958	Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
MYF5	99.99 %	159990	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155 (3), Autosomal recessive
MYH1	100 %	160730	*No OMIM phenotypes*
MYH14	99.98 %	608568	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant; Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant
MYH2	99.98 %	160740	Congenital myopathy 6 with ophthalmoplegia, 605637 (3), Autosomal dominant, Autosomal recessive
MYH3	99.99 %	160720	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant; 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
MYH7	99.99 %	160760	Laing distal myopathy, 160500 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Congenital myopathy 7B, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Congenital myopathy 7A, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant
MYH8	100 %	160741	Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant
MYL1	99.75 %	160780	Congenital myopathy 14, 618414 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MYL2	99.99 %	160781	Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant; Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424 (3), Autosomal recessive
MYLPF	99.85 %	617378	Arthrogryposis, distal, type 1C, 619110 (3), Autosomal dominant, Autosomal recessive
MYMK	99.88 %	615345	Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive
MYMX	100 %	619912	?Carey-Fineman-Ziter syndrome 2, 619941 (3), Autosomal recessive
MYO18B	99.98 %	607295	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive
MYO9A	99.93 %	604875	Myasthenic syndrome, congenital, 24, presynaptic, 618198 (3), Autosomal recessive
MYOD1	100 %	159970	Congenital myopathy 17, 618975 (3), Autosomal recessive
MYOT	99.93 %	604103	Myopathy, myofibrillar, 3, 609200 (3), Autosomal dominant
MYPN	99.88 %	608517	Cardiomyopathy, hypertrophic, 22, 615248 (3), Autosomal dominant; Congenital myopathy 24, 617336 (3), Autosomal recessive; Cardiomyopathy, familial restrictive, 4, 615248 (3), Autosomal dominant; Cardiomyopathy, dilated, 1KK, 615248 (3), Autosomal dominant
NEB	87.33 %	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive; Arthrogryposis multiplex congenita 6, 619334 (3), Autosomal recessive
NEFL	100 %	162280	Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
NFIX	99.99 %	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant
NPL	99.59 %	611412	*No OMIM phenotypes*
NRCAM	99.82 %	601581	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 (3), Autosomal recessive
NSUN3	99.94 %	617491	Combined oxidative phosphorylation deficiency 48, 619012 (3), Autosomal recessive
NUP88	99.96 %	602552	Fetal akinesia deformation sequence 4, 618393 (3), Autosomal recessive
OBSCN	99.99 %	608616	{Rhabdomyolysis, susceptibility to, 1}, 620235 (3), Autosomal recessive
ORAI1	99.63 %	610277	Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant
P2RX6	99.92 %	608077	*No OMIM phenotypes*
PABPN1	99.99 %	602279	Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant
PACSIN3	99.92 %	606513	*No OMIM phenotypes*
PAX7	99.83 %	167410	Congenital myopathy 19, 618578 (3), Autosomal recessive; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
PEX6	99.99 %	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal dominant, Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
PFKM	99.57 %	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
PGAM2	100 %	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive
PGK1	99.93 %	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM1	96.77 %	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
PHKA1	99.84 %	311870	Muscle glycogenosis, 300559 (3), X-linked recessive
PHKB	99.69 %	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PIEZO2	98.45 %	613629	Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant
PLEC	100 %	601282	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 (3), Autosomal recessive; Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex 5A, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive
PLEKHG5	99.99 %	611101	Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive
PNPLA2	100 %	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
PNPLA8	99.92 %	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
POGLUT1	99.99 %	615618	Dowling-Degos disease 4, 615696 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive
POLG	100 %	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLG2	99.51 %	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; ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 (3), Autosomal recessive
POLRMT	99.99 %	601778	Combined oxidative phosphorylation deficiency 55, 619743 (3), Autosomal dominant, Autosomal recessive
POMGNT1	99.69 %	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (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
POMGNT2	100 %	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
POMK	100 %	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
POMT1	99.96 %	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 (3), Autosomal recessive
POMT2	99.98 %	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 impaired intellectual development), type B, 2, 613156 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
POPDC3	99.96 %	605824	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848 (3), Autosomal recessive
PREPL	99.63 %	609557	Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
PRKAG2	99.96 %	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
PRUNE1	99.85 %	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
PTPN11	99.98 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTRH2	99.99 %	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
PUS1	100 %	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PYGM	99.96 %	608455	McArdle disease, 232600 (3), Autosomal recessive
PYROXD1	99.77 %	617220	Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive
RAPSN	99.97 %	601592	Fetal akinesia deformation sequence 2, 618388 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive
RBCK1	100 %	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
RBM7	99.99 %	612413	*No OMIM phenotypes*
REEP1	99.97 %	609139	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011 (3), Autosomal recessive; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751 (3), Autosomal dominant
RRM2B	99.97 %	604712	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
RXYLT1	99.48 %	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
RYR1	99.97 %	180901	Congenital myopathy 1B, autosomal recessive, 255320 (3), Autosomal recessive; Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000 (3), Autosomal dominant; King-Denborough syndrome, 619542 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 1}, 145600 (3), Autosomal dominant
RYR3	99.98 %	180903	Congenital myopathy 20, 620310 (3), Autosomal recessive
SCN4A	99.98 %	603967	Paramyotonia congenita, 168300 (3), Autosomal dominant; Hyperkalemic periodic paralysis, 170500 (3), Autosomal dominant; Congenital myopathy 22B, severe fetal, 620369 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Congenital myopathy 22A, classic, 620351 (3), Autosomal recessive
SELENON	93.61 %	606210	Congenital myopathy 3 with rigid spine, 602771 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SGCA	100 %	600119	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 (3), Autosomal recessive
SGCB	99.95 %	600900	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 (3), Autosomal recessive
SGCD	100 %	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 (3), Autosomal recessive
SGCG	99.99 %	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal recessive
SIGMAR1	99.99 %	601978	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726 (3), Autosomal recessive; ?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive
SIL1	99.95 %	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SLC16A1	99.26 %	600682	Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal dominant, Autosomal recessive
SLC18A3	99.99 %	600336	Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive
SLC22A5	99.99 %	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive
SLC25A1	99.93 %	190315	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive
SLC25A20	100 %	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
SLC25A26	99.76 %	611037	Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive
SLC25A4	100 %	103220	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant
SLC25A42	99.99 %	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive
SLC25A46	99.88 %	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
SLC52A2	100 %	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
SLC52A3	99.94 %	613350	?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive
SLC5A7	99.57 %	608761	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive
SMCHD1	99.83 %	614982	Facioscapulohumeral muscular dystrophy 2, digenic, 158901 (3), Digenic dominant; Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant
SMPX	99.8 %	300226	Myopathy, distal, 7, adult-onset, X-linked, 301075 (3), X-linked recessive; Deafness, X-linked 4, 300066 (3), X-linked dominant
SNAP25	99.89 %	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
SNUPN	99.99 %	607902	Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793 (3), Autosomal recessive
SORD	85.52 %	182500	Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912 (3), Autosomal recessive
SPEG	99.99 %	615950	Centronuclear myopathy 5, 615959 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SPTAN1	99.96 %	182810	Developmental delay with or without epilepsy, 620540 (3), Autosomal dominant; Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant; Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528 (3), Autosomal dominant
SPTBN4	99.91 %	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
SQSTM1	100 %	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
SRPK3	99.98 %	301002	*No OMIM phenotypes*
STAC3	99.91 %	615521	Congenital myopathy 13, 255995 (3), Autosomal recessive
STIM1	99.99 %	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
STT3A	99.99 %	601134	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 (3), Autosomal dominant; Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 (3), Autosomal recessive
SUCLA2	99.96 %	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SVIL	99.98 %	604126	Myofibrillar myopathy 10, 619040 (3), Autosomal recessive
SYNE1	99.95 %	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
SYNE2	99.95 %	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant
SYT2	99.93 %	600104	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 (3), Autosomal dominant; Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461 (3), Autosomal recessive
TAFAZZIN	99.98 %	300394	Barth syndrome, 302060 (3), X-linked recessive
TAMM41	99.97 %	614948	Combined oxidative phosphorylation deficiency 56, 620139 (3), Autosomal recessive
TANGO2	99.85 %	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TARDBP	100 %	605078	Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant; Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant
TBK1	99.07 %	604834	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant; Autoinflammation with arthritis and vasculitis, 620880 (3), Autosomal recessive
TCAP	100 %	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive
TEFM	99.88 %	616422	Combined oxidative phosphorylation deficiency 58, 620451 (3), Autosomal recessive
TFAM	99.24 %	600438	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 (3), Autosomal recessive
THOC2	99.11 %	300395	Intellectual developmental disorder, X-linked 12, 300957 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TIA1	99.72 %	603518	Welander distal myopathy, 604454 (3), Autosomal dominant, Autosomal recessive; Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133 (3), Autosomal dominant
TIMM22	100 %	607251	?Combined oxidative phosphorylation deficiency 43, 618851 (3), Autosomal recessive
TK2	99.96 %	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
TMEM126B	99.77 %	615533	Mitochondrial complex I deficiency, nuclear type 29, 618250 (3), Autosomal recessive
TMEM43	99.96 %	612048	Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Auditory neuropathy, autosomal dominant 3, 619832 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant
TMEM65	99.93 %	616609	*No OMIM phenotypes*
TNNC2	99.99 %	191039	Congenital myopathy 15, 620161 (3), Autosomal dominant
TNNI1	99.96 %	191042	*No OMIM phenotypes*
TNNI2	99.99 %	191043	Arthrogryposis, distal, type 2B1, 601680 (3), Autosomal dominant
TNNT1	99.82 %	191041	Nemaline myopathy 5C, autosomal dominant, 620389 (3), Autosomal dominant; Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355 (3), Autosomal recessive; Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386 (3), Autosomal recessive
TNNT3	100 %	600692	Arthrogryposis, distal, type 2B2, 618435 (3), Autosomal dominant
TNPO3	99.96 %	610032	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423 (3), Autosomal dominant
TNXB	90.86 %	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
TOP3A	99.91 %	601243	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 (3), Autosomal recessive
TOR1AIP1	98.45 %	614512	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 (3), Autosomal recessive
TPI1	99.95 %	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
TPM2	100 %	190990	Arthrogryposis, distal, type 2B4, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 1A, 108120 (3), Autosomal dominant; Congenital myopathy 23, 609285 (3), Autosomal dominant
TPM3	83.21 %	191030	Congenital myopathy 4A, autosomal dominant, 255310 (3), Autosomal dominant; Congenital myopathy 4B, autosomal recessive, 609284 (3), Autosomal recessive
TRAPPC11	99.93 %	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
TRAPPC2L	100 %	610970	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive
TRDN	99.8 %	603283	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441 (3), Autosomal recessive
TRIM32	100 %	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRIM54	89.88 %	606474	*No OMIM phenotypes*
TRIM63	99.96 %	606131	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TRIP4	99.97 %	604501	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive
TRMT5	99.99 %	611023	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539 (3), Autosomal recessive
TRPV4	100 %	605427	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); SED, Maroteaux type, 184095 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant
TSEN54	100 %	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
TSFM	100 %	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
TTN	99.15 %	188840	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 (3), Autosomal recessive; Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Congenital myopathy 5 with cardiomyopathy, 611705 (3), Autosomal recessive; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant; Cardiomyopathy, dilated, 1G, 604145 (3), Autosomal dominant; Myopathy, myofibrillar, 9, with early respiratory failure, 603689 (3), Autosomal dominant
TUBA4A	100 %	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant
TWNK	100 %	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
TYMP	99.87 %	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
UBA1	99.93 %	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3)
UNC45B	100 %	611220	?Cataract 43, 616279 (3), Autosomal dominant; Myofibrillar myopathy 11, 619178 (3), Autosomal recessive
VAMP1	100 %	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
VAPB	100 %	605704	Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant
VCP	99.99 %	601023	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3), Autosomal dominant; 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
VMA21	99.9 %	300913	Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive
VPS33B	99.95 %	608552	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
VRK1	99.98 %	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 (3), Autosomal recessive
VWA1	99.99 %	611901	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216 (3), Autosomal recessive
WARS1	99.97 %	191050	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317 (3), Autosomal recessive
YARS2	99.93 %	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive
ZBTB42	100 %	613915	?Lethal congenital contracture syndrome 6, 616248 (3), Autosomal recessive
ZC4H2	99.98 %	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.