The 2018 version of the gene table of monogenic neuromuscular disorders (nuclear genome)

Gisèle Bonne, Francois Rivier, Dalil Hamroun

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General features

This table is published annually in the December issue. Its purpose is to provide the reader of *Neuromuscular Disorders* with an updated list of monogenic muscle diseases due to a primary defect residing in the nuclear genome. It comprises diseases in which the causative gene is known or at least localized on a chromosome, if not yet identified. Diseases for which the locus has not been mapped or which are due to defects involving mitochondrial genes are not included.[1]

As in past years the diseases are classified into 16 groups:

1. *Muscular dystrophies;*
2. *Congenital muscular dystrophies;*
3. *Congenital myopathies;*
4. *Distal myopathies;*
5. *Other myopathies;*
6. *Myotonic syndromes;*
7. *Ion channel muscle diseases;*
8. *Malignant hyperthermias;*
9. *Metabolic myopathies;*
10. *Hereditary cardiomyopathies, subdivided into*
    10A *(non-arrhythmogenic) and*
    10B *(arrhythmogenic);*
11. *Congenital myasthenic syndromes;*
12. *Motor neurone diseases;*
13. *Hereditary ataxias;*
14. *Hereditary motor and sensory neuropathies;*
15. *Hereditary paraplegias;*
16. *Other neuromuscular disorders.*

In each group every entry corresponds to a clinical entity and has an item number.[2] A given gene may be involved in several different clinical entities (phenotypic heterogeneity such as in *LMNA* defects) and conversely a given clinical entity may be produced by a defect in several possible alternative genes (genotypic heterogeneity such as in CMT). In some diseases both kinds of heterogeneity may occur. As a consequence a gene or a disease may be cited in several places of the table.

The two versions of the gene table[3]

The **annual printed version** below is abridged and does not contain the *Arrhythmogenic Hereditary Cardiomyopathies (Group 10-B), Hereditary Ataxias (Group 13), and Hereditary Paraplegias (Group 15)*. The list of references is restricted to new key references corresponding to the items added or implemented since the preceding year.

The **full online version** contains the complete data of the 16 groups and the cumulative list of key references since 1991. It is freely accessible at http://www.musclegenetable.fr. It is designed to cope with the complexity described above. In this version the data are cross-referenced and linked to *PubMed* and to major databases related to molecular medicine *(Leiden Muscular Dystrophy, OMIM, NCBI, Genatlas, Orphanet, GeneCards)*. It contains several query tools allowing one to perform a variety of interrogations. This computerized version of the table is now surpassing the printed version which cannot accommodate the ever increasing volume and complexity of data. The **statistics tool** instantly provides the latest list of genes, proteins, phenotypes and cumulative bibliographic key references. Each list can be displayed, printed and exported in Excel format.

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[1] For diseases caused by mitochondrial genome mutations see: MITOMAP A human mitochondrial genome database. A compendium of polymorphisms and mutations of the human mitochondrial DNA http://www.mitomap.org/ MITOMAP.

[2] The assigned item number is provisional and may change in the next annual version.

[3] The history and development of both versions of the table are presented in the 2013 publication *(Kaplan JC and Hamroun D. The 2013 version of the gene table of neuromuscular disorders. Neuromuscul Disord. 22 (12), 1108–1135.)*

Overview of the new data in the 2018 printed version of the gene table

There are 35 new items, marked by background shading. Altogether they comprise **28 additional genes** and **8 additional phenotypic variants** caused by a gene already listed in the 2017 version (see box).

New in the 2017 printed version of the gene table
28 genes added:

AIFM1 (item # 16.34)
CALR3 (item # 10.20)
CNTNAP1 (item # 16.15)
DSG2 (item # 10.59)
ERBB4 (item # 12.57)
FASTKD2 (item # 16.36)
FLAD1 (item # 9.30)
HRAS (item # 3.50)
INPP5K (item # 2.49)
LAMA5 (item # 11.35)
MYMK (item # 3.51)
MYO18B (item # 3.49)
MYPN (item # 3.11 and item # 10.23)
NEFH (item # 14.66)
PMP2 (item # 14. 11)
POGLUT1 (item # 1.46)
PRDM1 (item # 10.68)
PTRH2 (item # 16.37)
RAF1 (item # 10.70)
SGPL1 (item # 14.80)
SLC18A3 (item # 11.27)
SLC25A46 (item # 12.73)
SPTBN4 (item # 3.47)
TMEM65 (item # 16.35)
TUBA4A (item # 12.60)
VAMP1 (item # 11.34 and item #15.66)
WARS (item # 12.23)
ZAK (item # 3.48)

8 additional phenotypic variants caused by mutation in a gene already listed in the gene table

SMCHD1
(item # 1.11 allelic to BAMS (OMIM #603457))
DAG1 (item # 2.37)
GYG1 (item # 9.12)
LDB3 (item # 10.25)
TCAP (item # 10.26)
BAG3 (item # 10.64 and 14.67)
VCP (item # 14.61)

1 new gene for a previously identified locus

SIGMAR1 (item #12.6, allelic to item # 12.54)

40 new key references

Contact

Users of the gene table are kindly requested to send any comments on the printed and/or the online version to g.bonne@institut-myologie.org.

Acknowledgements

We are extremely thankful to Jean-Claude Kaplan for his constant trust and support in giving us the opportunity to take over the maintenance of the "Muscle Gene Table" he initiated in 1991. We sincerely wish him an enjoyable retirement from the Gene Table, knowing he will keep a kindly eye on it. We sincerely thank Tanya Stojkovic for her careful review of entries in group 14.

We acknowledge the help of Myobase, a bibliographic alert system of the AFM (Asscociation Française contre les Myopathies), URL: http://www.myobase.org/

We are extremely appreciative of the invaluable assistance provided by Jane Miller at all stages of elaboration and editing of this table.

Gene table of monogenic neuromuscular disorders (nuclear genome only)

A computerized version of the table is freely accessible at *http://www.musclegenetable.fr/*

Shaded background indicates newly added items.

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
GROUP 1. MUSCULAR DYSTROPHIES								
Duchenne muscular dystrophy; Becker muscular dystrophy	1.1	XR	DMD 310200 BMD 300376	Xp21.2	***DMD*** *300377*	Dystrophin	Monaco et al. (1986) Burghes et al. (1987) Koenig et al. (1987, 1988) Hoffman et al. (1987, 1988)	Allelic to CMD3B (group 10)
Emery-Dreifuss muscular dystrophy, X-linked, type 1	1.2	XR	EDMD1 310300	Xq28	***EMD*** *300384*	Emerin	Hodgson et al. (1986) Romeo et al. (1988) Bione et al. (1994, 1995) Klauck et al. (1995) Nigro et al. (1995)	
Emery-Dreifuss muscular dystrophy, X-linked, type 2	1.3	XR	EDMD6 300696	Xq27.2	***FHL1*** *300163*	Four and a half LIM domain 1	Gueneau et al. (2009)	Allelic to RSS (2), XPMA (5), XPMD (5), reducing body myopathy (group 5)
Emery-Dreifuss muscular dystrophy, autosomal dominant	1.4	AD	EDMD2 181350	1q21.2	***LMNA*** *150330*	Lamin A/C	Bonne et al. (1999) Worman and Bonne (2007)	Allelic to EDMD3 (group 1), LGMD1B (group1), CMD1A (group 10), CMT2B1 (group 14), [+ several other phenotypes not in this table: FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, MADA/248370]
Emery-Dreifuss muscular dystrophy, autosomal recessive	1.5	AR	EDMD3 181350	1q21.2	***LMNA*** *150330*	Lamin A/C	Raffaele di Barletta et al. (2000) Worman and Bonne (2007)	Allelic to EDMD2 (group 1), LGMD1B (group 1), CMD1A (group 10), CMT2B1 (group 14), [+ several other phenotypes not in this table FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, not in this table]
Nesprin-1 related muscular dystrophy	1.6	AD	EDMD4 612998	6q25	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Zhang et al. (2007)	Allelic to dilated cardiomyopathy with nesprin-1 defect (group 10), SCAR8 (group 13), AMC with nesprin-1 defect (group 16)
Nesprin-2 related muscular dystrophy	1.7	AD	EDMD5 612999	14q23	***SYNE2*** *608442*	Spectrin repeat containing, nuclear envelope 2 (nesprin-2)	Zhang et al. (2007)	
LUMA-related muscular dystrophy	1.8	AD	EDMD7 614302	3p25.1	***(=LUMA)*** *612048*	Transmembrane protein 43 (=LUMA)	Liang et al. (2011)	Allelic to ARVD5 (group 10)
LAP1B-related muscular dystrophy	1.9	AR		1q25.2	***TOR1AIP1 (=LAP1B)*** *614512*	Torsin A interacting protein 1 (=Lamin-Associated Peptide 1B)	Kayman-Kurekci et al. (2014)	

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Facioscapulohumeral muscular dystrophy, type 1	1.10	AD	FSHMD1A 158900	4q35	***DUX4**** *606009* *(*inappropriate reactivation)*	Double homeobox 4	Wijmenga et al. (1990, 1991, 1992, 1993) Upadhyaya et al. (1990, 1992) Wright et al. (1993), van Deutekom et al. (1993) Gabellini et al. (2002), Van der Maarel et al. (2005) Gabellini et al. (2006) Petrov et al. (2006) Lemmers et al. (2010)	
Facioscapulohumeral muscular dystrophy, type 2	1.11	AD	FSHMD1B 158901	18p11.32	***SMCHD1**** ***(=KIAA0650)*** *614982* (*causing inappropriate reactivation of ***DUX4**** *606009*)	Structural maintenance of chromosomes flexible hinge domain containing 1	de Greef et al. (2010) Sacconi et al. (2012) Lemmers et al. (2012) Sacconi et al. (2013)	Allelic to Bosma Arhinia Microphthalmia Syndrome; BAMS (#603457)
Muscular dystrophy with generalized lipodystrophy	1.12	AD		17q21-q23	***PTRF*** *603198*	Polymerase I and transcript release factor (cavin-1)	Hayashi et al. (2009)	
Limb girdle muscular dystrophies, dominant								
LGMD1A	1.13	AD	LGMD1A 159000	5q31	***MYOT*** *604103*	Myotilin (titin immunoglobulin domain protein)	Speer et al. (1992) Hauser et al. (2000)	Allelic to distal myotilinopathy (group 4), MFM (group 5), spheroid body myopathy (group 5)
LGMD1B	1.14	AD	LGMD1B 159001	1q21.2	***LMNA*** *150330*	Lamin A/C	van der Koo et al (1997) Muchir et al. (2000) Worman and Bonne (2007)	Allelic to EDMD2 (group1), EDMD3 (group1), CMD1A (group10), CMT2B1 (group14), [+ several other phenotypes FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, not in this table]
LGMD1C	1.15	AD	LGMD1C 607801	3p25	***CAV3*** *601253*	Caveolin-3	Minetti et al. (1998) McNally et al. (1998)	Allelic to distal myopathy (group 4), hyper CKemia (group 5), RMD2 (group 6), CMH (group 10), LQT9 (group 10).
LGMD1D	1.16	AD	LGMD1D 603511	7q36	***DNAJB6*** *611332*	Hsp40 homolog, subfamily B, number 6	Speer et al. (1999), Sarparanta et al (2012) Harms et al. (2012)	
LGMD1E	1.17	AD	LGMD1E 602067	6q23	***DES*** *125660*	Desmin	Messina et al. (1997) Greenberg et al. (2012) Hedberg et al. (2012)	Allelic to myofibrillar myopathy (group 5) and CMD1I (group 10A), ARVD7 (group 10B)
LGMD1F	1.18	AD	LGMD1F 608423	7q32.1-q32.2	***TNPO3*** *610032*	Transportin 3	Palenzuela et al. (2003) Melià et al. (2013) Torella et al. (2013)	
LGMD1G	1.19	AD	LGMD1G 609115	4q21	***HNRPDL*** *607137*	Heterogeneous nuclear ribonucleoprotein D-like	Starling et al. (2005) Vieira et al. (2014)	
LGMD1H	1.20	AD	LGMD1H 613530	3p25.1-p23	*?*		Bisceglia et al. (2010)	
Limb girdle muscular dystrophies, recessive								
LGMD2A	1.21	AR	LGMD2A 253600	15q15.1	***CAPN3*** *114240*	calpain-3	Beckmann et al. (1991) Young et al. (1992), Richard et al. (1995, 1997)	
LGMD2B	1.22	AR	LGMD2B 253601	2p13.2	***DYSF*** *603009*	dysferlin	Bashir et al. (1994) Bashir et al. (1998) Liu et al. (1998)	Allelic to MM (group 4)
LGMD2C	1.23	AR	LGMD2C 253700	13q12	***SGCG*** *608896*	Gamma-sarcoglycan	Ben Othmane et al. (1992) Azibi et al. (1993) Noguchi et al. (1995) McNally et al. (1996) Piccolo et al. (1996)	

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LGMD2D	1.24	AR	LGMD2D 608099	17q21.33	***SGCA*** *600119*	alpha-sarcoglycan	Roberds et al. (1994) Piccolo et al. (1995) Passos-Bueno et al. (1995) Ljunggren et al. (1995) Carrié et al. (1997)	
LGMD2E	1.25	AR	LGMD2E 604286	4q12	***SGCB*** *600900*	beta-sarcoglycan	Lim et al. (1995) Bönnemann et al. (1995) Bönnemann et al. (1996)	
LGMD2F	1.26	AR	LGMD2F 601287	5q33.3	***SGCD*** *601411*	delta-sarcoglycan	Passos-Bueno et al. (1996) Nigro et al. (1996)	Allelic to CMD1L (group 10)
LGMD2G	1.27	AR	LGMD2G 601954	17q12	***TCAP*** *604488*	titin-cap (telethonin)	Moreira et al. (1997) Moreira et al. (2000)	Allelic to congenital muscular dystrophy with telethonin defect (group 2), CMD1N (group10)
LGMD2H	1.28	AR	LGMD2H 254110	9q31.2	***TRIM32*** *602290*	tripartite motif-containing 32	Weiler et al. (1998) Frosk et al. (2002)	Allelic to sarcotubular myopathy (group 3)
LGMD2I	1.29	AR	LGMD2I (MDDGC5) 607155	19q13.3	***FKRP*** *606596*	fukutin-related protein	Driss et al. (2000) Brockington et al. (2001a)	Allelic to MDC1C (group 2), WWS (group 2), MEB (group 2)
LGMD2J	1.30	AR	LGMD2J 608807	2q31	***TTN*** *188840*	titin	Hackman et al. (2003)	Allelic to congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), HMERF (group 5), CMH9 (group10), CMD1G (group10)
LGMD2K	1.31	AR	LGMD2K (MDDGC1) 609308	9q34.13	***POMT1*** *607423*	protein O-mannosyltransferase 1	Balci et al. (2005) D'Amico et al. (2006)	Allelic to WWS (group 2)
LGMD2L	1.32	AR	LGMD2L 611307	11p14.3	***ANO5 (TMEM16E)*** *608662*	anoctamin 5	Jarry et al. (2007), Bolduc et al. (2008, 2010), Hicks et al. (2011)	Allelic to early onset calf distal myopathy (group 4)
LGMD2M	1.33	AR	LGMD2M (MDDGC4) 611588	9q31-q33	***FKTN*** *607440*	fukutin	Murakami et al. (2006) Godfrey et al. (2006)	Allelic to FCMD (group 2), WWS (group2) dilated cardiomyopathy (group10)
LGMD2N	1.34	AR	LGMD2N (MDDGC2) 613158	14q24.3	***POMT2*** *607439*	protein O-mannosyl transferase 2	Biancheri et al. (2007)	Allelic to WWS (group 2) and to MEB (group 2)
LGMD2O	1.35	AR	LGMD2O (MDDGC3) 613157	1p34.1	***POMGNT1*** *606822*	protein O-linked mannose beta1,2-N-acetylglucosaminyl transferase 1	Godfrey et al. (2007); Clement et al. (2008) Raducu et al. (2012)	Allelic to WWS (group 2) and to MEB (group 2)
LGMD2P	1.36	AR	MDDGC9 613818	3p21	***DAG1*** *128239*	Alpha-dystroglycan Dystroglycan 1 (dystrophin-associated glycoprotein 1)	Hara et al. (2011)	Allelic to MDDGA9 (group 2)
LGMD2Q	1.37	AR	LGMD2Q 613723	8q24.3	***PLEC*** *601282*	plectin	Gundesli et al. (2010)	Allelic to LGMD with ophthalmoplegia (group 1) MDEBS (group 5), and Myasthenic syndrome with plectin defect (group 11)
LGMD2R	1.38	AR	LGMD2R 615325	2q35	***DES*** *125660*	desmin	Cetin et al. (2013)	Allelic to other desminopathies (groups 1, 5 and 10)
LGMD2S	1.39	AR	LGMD2S 615356	4q35.1	***TRAPPC11*** *614138*	trafficking protein particle complex 11	Bögershausen et al. (2013)	Allelic to CMD related to TRAPPC11 (group 2)
LGMD2T	1.40	AR	MDDGC14 615352	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Carss et al. (2013), Cabrera-Serrano et al. (2015)	Allelic to MDDGA14, MDDGB14 (group 2)
LGMD2U	1.41	AR	MDDGC7 616052	7p21.2	***ISPD*** *614631*	Isoprenoid synthase domain containing	Tasca et al. (2013)	Allelic to WWS/MDDGA7 (group 2)
LGMD2V	1.42	AR		17q25	***GAA*** *606800*	Glucosidase alpha, acid	Preisler et al. (2013)	Allelic to Pompe disease (groups 9 and 10)
LGMD2W	1.43	AR	616827	2q14.3	***LIMS2 (=PINCH2)*** *607908*	LIM and senescent cell antigen-like domains 2	Chardon et al. (2015)	
LGMD2X	1.44	AR	LGMD2X 616812	6q21	***BVES (=POPDC1)*** *604577*	blood vessel epicardial substance	Schindler et al. (2016)	

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LGMD2Y	1.45	AR	LGMD2Y 617072	1q25.2	***TOR1AIP1*** *614512*	torsin 1A interacting protein 1 (=lamin-associated protein 1)	Kayman-Kurekci et al. (2014); Sewry et al. (2014)	
LGMD2Z	1.46	AR	LGMD2Z 617232	3q13.33	***POGLUT1*** *615618*	Protein O-Glucosyltransferase 1	Servian-Morilla et al. (2016)	
Muscle dystrophy with glycosylation defect, type Io	1.47	AR	CDG1O 612937	1q22	***DPM3*** *605951*	Dolichyl-phosphate mannosyltransferase polypeptide 3	Lefeber et al. (2009)	
Scapuloperoneal muscular dystrophy and dropped head syndrome	1.48	AR	600416	9p13-p12	***VCP*** *601023*	Valosin-containing protein	Liewluck et al. (2014)	Allelic to IBMPFD (groups 4 and 5), to ALS14 (group 12) and CMT2Y (group 14)
LGMD with ophthalmoplegia	1.49	AR		8q24.3	***PLEC*** *601282*	Plectin	Fattahi et al. (2015)	Allelic to LGMD2Q (group 1), MDEBS (group 5), myasthenic syndrome with plectin defect (group 11)
GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES								
Congenital muscular dystrophy with merosin deficiency	2.1	AR	MDC1A 607855	6q2	***LAMA2*** *156225*	Laminin alpha2 chain of merosin (=laminin-2)	Tomé et al. (1994) Hillaire et al. (1994) Helbling-Leclerc et al. (1995) Allamand et al. (1997)	
Bethlem myopathy	2.2	AD	BTHLM1 158810	21q22.3	***COL6A1*** *120220*	collagen type VI, subunit alpha 1	Jöbsis et al. (1996)	allelic to UCMD (group 2)
Bethlem myopathy	2.3	AD	BTHLM1 158810	21q22.3	***COL6A2*** *120240*	collagen type VI, subunit alpha 2	Jöbsis et al. (1996)	allelic to UCMD (group 2), and myosclerosis (group 2)
Bethlem myopathy	2.4	AD	BTHLM1 158810	2q37	***COL6A3*** *120250*	collagen type VI, subunit alpha 3	Speer et al. (1996) Bertini et al. (1998) Pan et al. (1998)	allelic to UCMD (group 2)
Bethlem myopathy (recessive)	2.5	AR	BTHLM1 158810	21q22.3	***COL6A2*** *120240*	collagen type VI, subunit alpha2	Gualandi et al. (2009)	allelic to UCMD (group 2)
Ullrich syndrome	2.6	AR	UCMD 254090	21q22.3	***COL6A1*** *120220*	collagen, type VI, subunit alpha 1	Pan et al. (2003) Giusti et al. (2005)	allelic to Bethlem myopathy (group 2)
Ullrich syndrome	2.7	AR	UCMD 254090	21q22.3	***COL6A2*** *120240*	collagen, type VI, subunit alpha 2	Vanegas et al. (2001) Higuchi et al. (2001)	allelic to Bethlem myopathy (group 2) and myosclerosis (group 2)
Ullrich syndrome	2.8	AR	UCMD 254090	2q37	***COL6A3*** *120250*	collagen type VI, subunit alpha 3	Demir et al. (2002)	allelic to allelic to Bethlem myopathy (group 2) and myosclerosis (group 2)
Ullrich congenital muscular dystrophy 2	2.9	AR	UCMD2 616470	*6q13-q14*	***COL12A1*** *120320*	collagen type XII, alpha 1 chain	*Zou et al. (2014)*	
Bethlem myopathy 2	2.10	AD	BTHLM2 *616471*	*6q13-q14*	***COL12A1*** *120320*	collagen type XII, alpha 1 chain	*Zou et al. (2014)*	
COL12A1-related congenital muscular dystrophy	2.11	AD		*6q13-q14*	***COL12A1*** *120320*	collagen type XII, alpha 1 chain	Punetha et al. *(2016)*	
Myosclerosis	2.12	AR	255600	21q22.3	***COL6A2*** *120240*	collagen type VI, subunit alpha 2	Merlini et al. (2008)	allelic to UCMD (group 2) and to Bethlem myopathy (group 2)
Rigid spine syndrome related to *SEPN1*	2.13	AR	RSMD1 602771	1p36	***SEPN1*** *606210*	selenoprotein N1	Moghadaszadeh et al. (1998, 2001) Ferreiro et al. (2002a, 2004)	allelic to CFTD (group 3), multiminicore disease (group 3), and desmin-related myopathy with Mallory bodies (group 5)
Rigid spine syndrome related to *FHL1*	2.14	AR	RSMD1 602771	Xq26.3	***FHL1*** *300163*	four and a half LIM domain 1	Shalaby et al. (2008	allelic to EDMD6 (1), RSS (2), XPMA(5), XPMD (5)
Congenital muscular dystrophy related to integrin	2.15	AR	613204	12q13	***ITGA7*** *600536*	integrin α7	Hayashi et al. (1998)	
Congenital muscular dystrophy related to dynamin 2	2.16	AD		19p13.2	***DNM2*** *602378*	dynamin 2	Susman et al. (2008)	allelic to CNM (group 3) and CMTIB (group 14)
Congenital muscular dystrophy related to telethonin	2.17	AR		17q12	***TCAP*** *604488*	titin-cap (telethonin)	Ferreiro et al. (2011)	Allelic to LGMD2G (group 1) and to CMD1N (group 10A)
Congenital muscular dystrophy related to *LMNA*	2.18	AD	MDCL 613205	1q21.2	***LMNA*** *150330*	Lamin A/C	Quijano-Roy et al. (2008)	Allelic to EDMD3 (group 1), LGMD1B (group 1), L-CMD (group 2), CMD1A (group 10/A), CMT2B1 (group 14), [+ several other phenotypes not in this table: FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, MADA/248370]

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Congenital muscle dystrophies due to defective glycosylation								
Fukuyama congenital muscular dystrophy	2.19	AR	MDDGA4 253800 MDDGB4 613152	9q31-q33	***FKTN*** *607440*	fukutin	Toda et al. (1993) Kobayashi et al. (1998)	allelic to WWS (group 2)
Walker-Warburg syndrome (WWS)	2.20	AR	MDDGA4 253800 MDDGB4 613152	9q31-q33	***FKTN*** *607440*	fukutin	Beltran-Valero de Bernabe (2003) Mercuri et al. (2009)	allelic to LGMD2L (group 1) and Fukuyama (group 2)
Walker-Warburg syndrome (WWS)	2.21	AR	MDDGA1 236670 MDDGB1 607423	9q34	***POMT1*** *607423*	protein-O-mannosyltransferase 1	Beltran-Valero De Bernabe et al. (2002) van Reeuwijk et al. (2006) Mercuri et al. (2009)	allelic to LGMD2K (group 1)
Walker-Warburg syndrome (WWS)	2.22	AR	MDDGA2 613150 MDDGB2 613156	14q24.3	***POMT2*** *607439*	protein O-mannosyl transferase 2	van Reeuwijk et al. (2005) Mercuri et al. (2009)	allelic to LGMD2N (group 1) and MEB (group 2)
Walker-Warburg syndrome (WWS)	2.23	AR	MDDGA5 613153 MDDGB5 606612	19q13	***FKRP*** *606596*	fukutin-related protein	Beltran-Valero De Bernabe et al. (2004)	allelic to LGMD2I (group 1), MDC1C (group 2), MEB (group 2)
Walker-Warburg syndrome (WWS)	2.24	AR	MDDGA3 253280 MDDGB3 613151	1p34.1	***POMGNT1*** *606822*	O-mannose beta1,2-N-acetylglucosaminyl transferase	Taniguchi et al. (2003) Mercuri et al. (2009)	allelic to MEB (group 2)
Walker-Warburg syndrome (WWS)	2.25	AR	MDDGA7 614643	7p21.2	***ISPD*** *614631*	isoprenoid synthase domain containing	Roscioli et al. (2012) Willer et al. (2012)	
Walker-Warburg syndrome (WWS)	2.26	AR	MDDGA8 614830	3p22.1	***GTDC2*** *614828*	glycosyltransferase-like domain containing 2	Manzini et al. (2012)	
Walker-Warburg syndrome (WWS)	2.27	AR	MDDGA13 615287	11q13.2	***B3GNT1*** *605517*	UDP-GlcNAc: betaGal beta-1,3-N-acetylglucosaminyl-transferase 1	Buysse et al. (2013) Shaheen et al. (2013	
Muscle-eye-brain disease (MEB)	2.28	AR	MDDGA3 253280	1p34.1	***POMGNT1*** *606822*	O-mannose beta1,2-N-acetylglucosaminyl transferase	Yoshida et al. (2001) Taniguchi et al. (2003)	allelic to WWS (group 2)
Muscle-eye-brain disease (MEB)	2.29	AR	MDDGA5 613153 MDDGB5 606612	19q13.32	***FKRP*** *606596*	Fukutin-related protein	Beltran-Valero De Bernabe et al. (2004)	allelic to LGMD2I (group 1), MDC1C/(group 2), WWS (group 2)
Muscle-eye-brain disease (MEB)	2.30	AR	MDDGA2 613150 MDDGB2 613156	14q24.3	***POMT2*** *607439*	protein O-mannosyl transferase 2	Mercuri et al. (2006)	allelic to WWS (group 2)
Muscle-eye-brain disease (MEB)	2.31	AR	MDDGC14 615350	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	de Carss et al. (2013)	Allelic to LGMD2T (group 1) and to MDDGB14 (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.32	AR	MDDGA5 613153 MDDGB5 606612	19q13	***FKRP*** *606596*	Fukutin-related protein	Brockington et al. (2001b) Topaloglu et al. (2003) Mercuri et al. (2009)	allelic to LGMD2I (group 1), WWS (group 2), MEB (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.33	AR	MDDGA6 608840 MDDGB6 613154	22q12	***LARGE*** *603590*	like-glycosyl transferase	Longman et al. (2003) Mercuri et al. (2009)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.34	AR	CDG1E 608799	20q13.13	***DPM1*** *603503*	dolichyl-phosphate mannosyltransferase 1, catalytic subunit	Yang et al. (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and severe epilepsy	2.35	AR	CDG1U 615042	9q34.13	***DPM2*** *603564*	dolichyl-phosphate mannosyltransferase polypeptide 2, regulatory subunit	Barone et al. (2012)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.36	XR	CDG1S 300884	Xq23	***ALG13*** *300776*	UDP-N-acetylglucosami-nyltransferase subunit	Timal et al. (2012	Congenital myasthenia Group 11.
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A9	2.37	AR	MDDGA9 616538	3p21	***DAG1*** *128239*	Dystrophin-Associated Glycoprotein 1	Geis et al. (2013)	Allelic to LGMD2P (group 1)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A10	2.38	AR	MDGGA10 615041	12q14.2	***TMEM5*** *605862*	transmembrane protein 5	Vuillaumier-Barrot et al. (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan WWWS/MEB like	2.39	AR	MDDGA11 615181	1q42.3	***B3GALNT2*** *610194*	beta-1,3-N-acetylgalactosaminyl-transferase 2	Stevens et al. (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A12	2.40	AR	MDDGA12 615249	8p11.21	***POMK*** *(=SGK196) 615247*	protein-O-mannose kinase	Jae et al. (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and mental retardation	2.41	AR	MDDGB14 615351	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Carss et al. (2013)	Allelic to LGMD2T (group 1), MDDGC14 (group 2)
Other congenital muscular dystrophies								
Congenital muscle dystrophy with joint hyperlaxity	2.42	AR		3p23-21	*?*		Tetreault et al. (2006)	
Congenital muscle dystrophy with mitochondrial structural abnormalities (megaconial type)	2.43	AR	MDCMC 602541	22q13	***CHKB*** *612395*	Choline kinase beta	Mitsuhashi et al. (2011)	
Congenital muscular dystrophy	2.44	AR	MDC1B 604801	1q42	*?*		Brockington et al. (2000)	
Congenital muscular dystrophy with rigid spine related to *ACTA1*	2.45	AR	Possibly identical to MDC1B 604801	1q42.1	***ACTA1*** *102610*	Alpha actin, skeletal muscle	O'Grady et al. (2014)	Allelic to NEM3, CFTD, cap myopathy related to ACTA1 (group 3)
Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by *TRAPPC11* mutations	2.46	AR	CMD	4q35.1	***TRAPPC11*** *614138*	Trafficking protein particle complex 11	Liang et al. (2015)	Allelic to LGMD2S (group 1)
GOLGA2-related congenital muscle dystrophy with brain involvement	2.47	AR		9q34.113	***GOLGA2*** *602580*	golgin A2	Shamseldin et al. (2016)	
Muscular dystrophy, congenital Davignon-Chauveau type	2.48	AR	MDCDC 617066	15q22.31	***TRIP4*** *604501*	thyroid hormone receptor interactor 4	Davignon et al. (2016)	allelic to SMABF1 (group 12)
Muscular dystrophy, congenital, with cataracts and intellectual disability	2.49	AR	MDCCAID 617404	17p13.3	***INPP5K*** *607875*	Inositol Polyphosphate-5-Phosphatase K	Osborn et al. (2017) Wiessner et al. (2017)	
GROUP 3. CONGENITAL MYOPATHIES								
Nemaline myopathy								
NEM1	3.1	AD	NEM1 609284	1q21.2	***TPM3*** *191030*	Tropomyosin 3	Laing et al. (1992) Laing et al. (1995b) Tan et al. (1999) Wattanasirichaigoon et al. (2002)	
NEM2	3.2	AR	NEM2 256030	2q22	***NEB*** *161650*	Nebulin	Wallgren-Pettersson et al. (1995, 2002) Pelin et al. (1999) Lehtokari et al. (2006)	
NEM3	3.3	AD	NEM3 161800	1q42.1	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Nowak et al. (1999)	allelic to CFTD (group 3)
NEM4	3.4	AD	NEM4 609285	9p13	***TPM2*** *190990*	Tropomyosin 2 (beta)	Donner et al. (2002) Monnier et al. (2009)	
NEM5	3.5	AR	NEM5 605355	19q13	***TNNT1*** *191041*	Troponin T type 1 (skeletal, slow)	Johnston et al. (2000	
NEM6	3.6	AD	NEM6 609273	15q22.31	***KBTBD13*** *613727*	Kelch repeat and BTB (POZ) domain containing 13	Gommans et al. (2003) Samburghin et al. (2010)	
NEM7	3.7	AR	NEM7 610687	14q12	***CFL2*** *601443*	Cofilin 2 (muscle)	Agrawal et al. (2007)	
NEM8	3.8	AR	NEM8 615348	2p22.1	***KLHL40*** *615340*	Kelch-like family member 40	Ravenscroft et al. (2013)	
NEM9	3.9	AR	NEM9 615731	2q31.1	***KLHL41*** *607701*	Kelch-like family member 41	Gupta et al. (2013)	
NEM10	3.10	AR	NEM 10 616165	3p14.1	***LMOD3*** *616112*	Leiomodin 3 (fetal)	Yuen et al. (2014)	
NEM11	3.11	AE	NEM11 617336	10q21.3	***MYPN*** *608517*	Myopalladin	Miyatake et al. (2016) Lornage et al. (2017)	Allelic to CMD1KK and CMD22 (group 10A)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Myopathy, congenital, with fiber-type disproportion	3.12	AD	CFTD 255310	1q42.1	***ACTA1*** *102610*	actin, alpha 1, skeletal muscle	Clarke et al. (2003); Laing et al. (2004)	allelic to NEM3 (group 3)
Myopathy, congenital, with fiber-type disproportion	3.13	AR	CFTD 255310	1p36	***SEPN1*** *606210*	selenoprotein N1	Clarke et al. (2006)	allelic to RSMD1 (group 2), multiminicore disease (group 3), desmin-related myopathy with Mallory bodies (group 5)
Myopathy, congenital, with fiber-type disproportion	3.14	AD	CFTD 255310	1q21.2	***TPM3*** *191030*	tropomyosin 3	Clarke et al. (2008)	allelic to NEM1 (group 3)
Myopathy, congenital, with fiber-type disproportion	3.15	AR	CFTD 255310	19q13.1	***RYR1*** *180901*	ryanodine receptor	Clarke et al. (2010)	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Myopathy congenital, with fiber-type disproportion	3.16	AD	CFTD 255310	14q12	***MYH7*** *160760*	myosin, heavy chain 7, cardiac muscle, b	Ortolano et al. (2011)	allelic to MPD1 (group 4), CMH1 (group 10), CMD1S (group 10)
Myotubular myopathy	3.17	XR	CNMX 310400	Xq28	***MTM1*** *300415*	myotubularin 1	Thomas et al. (1987); Laporte (1996, 1997, 2000)	
Centronuclear myopathy related to *DNM2*	3.18	AD	CNM1 160150	19.p13.2	***DNM2*** *602378*	dynamin 2	Bitoun et al. (2005)	
Centronuclear myopathy related to *BIN1*, recessive	3.19	AR	CMN2 255200	2q14	***BIN1*** *601248*	amphiphysin	Nicot et al. (2007) Böhm et al. (2014)	
Centronuclear myopathy related to *BIN1,* dominant	3.20	AD		2q14	***BIN1*** *601248*	amphiphysin	Böhm et al. (2014)	
Centronuclear myopathy related to *RYR1*	3.21	AR	255200	19q13.1	***RYR1*** *180901*	ryanodine receptor	Wilmshurst et al. (2010)	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Centronuclear myopathy related to *TTN*	3.22	AR		2q24.3	***TTN*** *188840*	titin	Ceyhan-Birsoy et al. (2013)	Allelic to # 1.29, 3.34, 4.2, 5.13, 10.8, 10.33
Centronuclear myopathy with dilated cardiomyopathy	3.23	AR	CMN5 615959	2q35	***SPEG*** *615950*	SPEG complex locus	Agrawal et al. (2014)	
Central core disease, dominant	3.24	AD	CCD 117000	19q13.1	***RYR1*** *180901*	ryanodine receptor	Kausch et al. (1991) Zhang et al. (1993) Quane et al. (1993), Robinson et al. (2002)	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Central core disease, recessive (transient multiminicore myopathy)	3.25	AR	CCD 117000	19q13.1	***RYR1*** *180901*	ryanodine receptor	Ferreiro et al. (2002a); Jungbluth et al. (2002)	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Multiminicore disease with external ophthalmoplegia	3.26	AR	255320	19q13.1	***RYR1*** *180901*	ryanodine receptor	Monnier et al. (2003), Jungbluth et al. (2005)	allelic to CCD (group 3), CCD (group 3), MHS1 (group 8)
Multiminicore disease, classical form	3.27	AR	255320	1p36	***SEPN1*** *606210*	selenoprotein N1	Ferreiro et al. (2002b), Ferreiro et al. (2004)	allelic to RSMD1 (group 2), desmin related myopathy with Mallory bodies (group 5)
Early-onset myopathy, areflexia, respiratory distress and dysphagia	3.28	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	multiple EGF-like domains 10	Logan et al. (2011)	See item # 3. 28
Recessive congenital myopathy with minicores	3.29	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	multiple EGF-like domains 10	Boyden et al. (2012)	See item # 3. 27
Hyaline body myopathy (recessive)	3.30	AR	255160	3p22.2-p21.32	*?*	?	Onengut et al. (2004)	
Hyaline body myopathy, dominant (myosin storage myopathy)	3.31	AD	MSMA 608358	14q12	***MYH7*** *160760*	myosin, heavy chain 7, cardiac muscle, b	Tajsharghi et al. (2003) Bohlega et al. (2004) Laing et al. (2005)	allelic to CFTD (group 3), MPD1 (group 4), CMH1 (group 10), CMD1S (group 10)
Myosin storage myopathy and cardiomyopathy, recessive	3.32	AR		14q12	***MYH7*** *160780*	myosin, heavy chain 7, cardiac muscle, b	Tajsharghi et al. (2007a)	allelic to CFTD (group 3), MPD1 (group 4), CMH1 (group 10), CMD1S (group 10)
Myosin IIa myopathy, dominant (Inclusion body myopathy 3)	3.33	AD	IBM3 605637	17p13.1	***MYH2*** *160740*	myosin, heavy chain 2, skeletal muscle, adult	Martinsson et al. (1999, 2000)	
Myopathy proximal with ophthalmoplegia dominant (Inclusion body myopathy 3)	3.34	AD	MYPOP 605637	17p13.1	***MYH2*** *160740*	myosin, heavy chain 2, skeletal muscle, adult	Martinsson et al. (1999, 2000)	
Myopathy proximal with ophthalmoplegia recessive (Inclusion body myopathy 3)	3.35	AR	MYPOP 605637	17p13.1	***MYH2*** *160740*	myosin, heavy chain 2, skeletal muscle, adult	Tajsharghi et al. (2010)	
Isolated inclusion body myopathy	3.36	AD	IBMPFD3 615424	12q13.13	***HNRPA1*** *164017*	heterogeneous nuclear ribonucleoprotein A1	Izumi et al. (2015)	allelic to ALS20 (group 12)

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Cap myopathy	3.37	AD	CAPM2 609285	9p13	***TPM2*** *190990*	tropomyosin 2, b	Tajsharghi et al. (2007b) Lehtokari et al. (2007)	allelic to NEM4 (group 3) DA1 (group 16) and DA2B (group 16)
Cap myopathy	3.38	AD	CAPM1 609284	1q21.2	***TPM3*** *191030*	tropomyosin 3	De Paula et al. (2009) Ohlsson et al. (2009)	allelic to NEM1 (group 3)
Cap myopathy	3.39	AD		1q42.1	***ACTA1*** *102610*	actin, alpha 1, skeletal muscle	Hung et al. (2010)	allelic to NEM3 (group 3)
Congenital neuromuscular disease with uniform type 1 fiber	3.40	AR, AD	CCD 117000	19q13.1	***RYR1*** *180901*	ryanodine receptor I	Sato et al. (2007)	allelic to CDD (group 3), multi-minicore disease (group 3), MHS1 (group 8)
Congenital myopathy with fatal cardiomyopathy	3.41			2q31	***TTN*** *188840*	titin	Carmignac et al. (2007)	allelic to LGMD2J (group 1), TMD (group 4,) HMERF (group 5), CMH9 (group 10), CMD1G (group 10)
Congenital skeletal myopathy and fatal cardiomyopathy	3.42	AR		11p11.2	***MYBPC3*** *600958*	cardiac myosin binding protein-C	Tajsharghi et al. (2010)	allelic to CMH4 (group 10)
Congenital lethal myopathy	3.43	AR	MYPCN 612540	12q11-q12	***CNTN1*** *600016*	contactin-1	Compton et al. (2008)	
Sarcotubular myopathy	3.44	AR		9q31	***TRIM32*** *602290*	tripartite motif containing 32 (ubiquitin ligase)	Schoser et al. (2005)	allelic to LGMD2H (group 1)
Congenital myopathy related to *PTPLA*	3.45	AR		10p12.33	**HACD1** (=PTPLA) *610467*	protein tyrosine phosphatase-like (3-Hydroxyacyl-CoA dehydratase)	Muhammad et al. (2013)	ARVD6 (item # 10–80)
Congenital myopathy with ophthalmoplegia related to *CACNA1S*	3.46	AR		1q32.1	***CACNA1S*** *114208*	calcium channel, voltage-dependent, L type, alpha 1S subunit	Hunter et al. (2015)	Allelic to hypoKPP1 (group 7) MHS5 (group 8)
Myopathy, Congenital, With Neuropathy And Deafness	3.47	AR	CMND 617519	19q13	***SPTBN4*** *606214*	Spectrin, Beta, Nonerythrocytic, 4	Knierim et al. (2017)	
Myopathy, Congenital, with fiber type disproportion	3.48	AR		2q31.1	***ZAK*** *609479*	Leucine Zipper-And Sterile Alpha Motif-Containing Kinase	Vasli et al. (2017)	
Nemaline Myopathy with Cardiomyopathy	3.49	AR		22q12.1	***MYO18B*** *607295*	Myosin XVIIIB	Malfatti et al. (2015)	
Myopathy, congenital, With excess of muscle spindles	3.50	AD	CMEMS 218040	11p15.5	***HRAS*** *190020*	V-Ha-RAS Harvey Rat Sarcoma Viral Oncogene Homolog	Quélin et al. (2017)	
Myopathy, Congenital nonprogressive, with Moebius sequence and Robin sequence	3.51	AR	CFZS 254940	9q34.2	***MYMK*** *615345*	Myomaker	Di Gioia et al. (2017)	
GROUP 4. DISTAL MYOPATHIES								
Distal recessive myopathy (Miyoshi)	4.1	AR	MMD1 254130	2p12-14	***DYSF*** *603009*	dysferlin	Bejaoui et al. (1995) Bashir et al. (1998) Liu et al. (1998)	allelic to LGMD2B (group 1)
Tibial muscular dystrophy (Udd)	4.2	AD	TMD 600334	2q31	***TTN*** *188840*	titin	Haravuori et al. (1998) Haravuori et al. (2001) Hackman et al. (2002)	allelic to LGMD2J (group 1),congenital myopathy with fatal cardiomyopathy (group 3), HMERF (group 5), CMH9 (group 10), CMD1G (group 10)
Distal myopathy with rimmed vacuoles (Nonaka) and Hereditary inclusion body myopathy	4.3	AR	NM 605820 IBM2 600737	9p12-p12	***GNE*** *603824*	glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase	Mitrani-Rosenbaum et al. (1996), Ikeuchi et al. (1997) Eisenberg et al. (2001)	
Distal myopathy (Laing)	4.4	AD	MPD1 160500	14q11.2	***MYH7*** *160760*	myosin, heavy chain 7, cardiac muscle, beta	Laing et al. (1995a), Mastaglia et al. (2000), Meredith et al. (2004)	allelic to CFTD (group 3), myosin storage myopathy (group 3), CMH1 (group 10), CMD1S (group 10)
Vocal cord and pharyngeal distal myopathy	4.5	AD	MPD2 606070	5q31	***MATR3*** *164015*	Matrin 3	Feit et al. (1998) Senderek et al. (2009)	Allelic to ALS21 (group 12)
Adult-onset distal myopathy	4.6	AD	MPD3 610099	8p22-q11	*?*		Haravuori et al. (2004)	
Welander distal myopathy	4.7	AD	WDM 604454	2p13	***TIA1*** *603518*	cytotoxic granule-associated RNA binding protein	Ahlberg et al. (1999) Hackman et al. (2013) Klar et al (2013)	

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Distal myopathy with pes cavus and areflexia (Vacuolar neuromyopathy)	4.8	AD	601846	19.p13	*?*		Servidei et al. (1999)	
Distal myopathy with myotilin defect	4.9	AD		5q31	***MYOT*** *604103*	myotilin	Penisson-Besnier et al. (1998, 2006)	allelic to LGMD1A (group 1), MFM (group 5), spheroid body myopathy (group 5)
Distal myopathy with nebulin defect	4.10	AR		2q22	***NEB*** *161650*	nebulin	Wallgren-Pettersson et al. (2007)	allelic to NEM2 (group 3)
Distal myopathy with caveolin defect	4.11	AD		3p25	***CAV3*** *601253*	caveolin-3	Tateyama et al. (2002); Fulizio et al. (2005)	allelic to LGMD1C (group 1), hyper CKemia (group 5), RMD2 (group 6), CMH (group 10)
Late-onset distal myopathy (Markesbery-Griggs)	4.12	AD		10q23.2	***LDB3*** *(=ZASP) 605906*	LIM domain binding -3 (Z band alternatively spliced PDZ motif)	Griggs et al. (2007)	allelic to MFM (group 5)
Early-onset calf distal myopathy	4.13	AR	MMD3 613319	11p14-12	***AN05*** *608662*	anoctamin 5	Bolduc et al. (2010)	allelic to LGMD2L (group 1)
Dynamin 2-related distal myopathy	4.14	AD	CNM1 160150	19p13.2	***DNM2*** *602378*	dynamin 2	Fischer et al (2006)	allelic to CNM (group 3) and CMTDIB (group 14)
Early-onset distal myopathy with KLHL9 defect	4.15	AD		9p22	***KLHL9*** *611201*	Kelch-like homolog 9	Cirak et al. (2010)	
Filamin C-related distal myopathy	4.16	AD	MPD4 614065	7q32	***FLNC*** *102565*	filamin C, gamma (actin binding protein 280)	Duff et al. (2011)	allelic to MFM5 (group 5)
Distal myopathy with VCP defect	4.17	AD	IBMPFD 167320	9p13-p12	***VCP*** *601023*	valosin-containing protein	Palmio et al. (2011)	Allelic to #1.48, IBMBFD (group 5), ALS14 (group 12) and CMT2Y (group 14)
Adolescent-onset distal myopathy	4.18	AR	MPD5 617030	14q32-33	***ADSSL1*** *612498*	Adénylosuccinate synthase-like	Park et al. (2018)	
GROUP 5. OTHER MYOPATHIES								
A. Myofibrillar myopathies								
Myofibrillar myopathy, desmin-related myopathy	5.1	AD	MFM1 601419	2q35	***DES*** *125660*	desmin	Goldfarb et al. (1998) Munoz-Marmol et al. (1998)	Allelic to LGMD1E (group 1), CMD1I (group 10/A), ARVD7 (group 10/B)
Myofibrillar myopathy, alpha-B crystallin related	5.2	AD	MFM2 608810	11q22	***CRYAB*** *123590*	crystallin, alpha B	Vicart et al. (1998); Selcen et al. (2003)	
Myofibrillar myopathy, myotilin related	5.3	AD	MFM3 609200	5q31	***MYOT*** *604103*	myotilin (titin immunoglobulin domain protein)	Selcen and Engel (2004)	allelic to LGMD1A (group 1), spheroid body myopathy (group 5)
Spheroid body myopathy	5.4	AD	MFM3 182920	5q31	***MYOT*** *604103*	myotilin (titin immunoglobulin domain protein)	Foroud et al. (2005)	allelic to LGMD1A (group 1), MFM (group 5)
Myofibrillar myopathy	5.5	AD	MFM4 609452	10q22	***LDB3*** *= ZASP 605906*	LIM domain binding-3 (Z band alternatively spliced PDZ motif)	Selcen and Engel (2005)	allelic to Markesbery-Griggs (group 4) and to CMD1C (group 10)
Myofibrillar myopathy, filamin-C related	5.6	AD	MFM5 609524	7q32	***FLNC*** *102565*	filamin C, gamma (actin binding protein 280)	Vorgerd et al. (2005)	Allelic to filamin C related distal myopathy (group 4)
Myofibrillar myopathy with BAG3 defect	5.7	AD	MFM6 612954	10q25-q26	***BAG3*** *603883*	BCL2-associated athanogene 3	Selcen et al. (2009)	
Myopathy microfibrillar type 7	5.8	AR	MFM7 617114	3q22.2	***KY*** *605739*	Kyphoscoliosis peptidase	Hedberg-Oldfors et al. (2016); Straussberg et al. (2016)	
Early-onset myofibrillar myopathy with PYRODX1 defect	5.9	AR	MFM8 617258	12p12.1	***PYRODX1*** *617220*	Pyridine nucleotide-disulphide oxidoreductase domain 1	O'Grady et al. (2016)	
Desmin-related myopathy with Mallory bodies	5.10	AD	602771	1p36	***SEPN1*** *606210*	selenoprotein N1	Ferreiro et al. (2004)	allelic to RSMD1 (group 2), CFTD (group 3) multiminicore disease (group 3)
Cardiac and skeletal aggregate myopathy	5.11	Digenic		1p36.11 2p.23.3	***TRIM63*** *(=MURF1) 606131* ***+ TRIM54*** *(=MURF3) 606474*	tripartite motif containing 63, E3 ubiquitin protein ligase + tripartite motif containing 54	Olivé et al. (2015)	

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Myofibrillar myopathy with arrhythmogenic right ventricular cardiomyopathy	5.12	AD	MFM/ARVC 609160	10q22	*?*		Melberg et al. (1999) Kuhl et al. (2008)	
B. Miscellaneous								
Danon disease	5.13	XD	GSD IIb 300257	Xq24	***LAMP2*** *309060*	lysosomal-associated membrane protein 2	Nishino et al. (2000) Musumeci et al. (2005)	
Myopathy with excessive autophagia	5.14	XR	MEAX (XMEA) 310440	Xq28	***VMA21*** *300913*	VMA21 Vacuolar H+-ATPase Homolog (S. cerevisiae)	Saviranta et al. (1988) Villard et al. (2000) Minassian et al. (2002); Munteanu et al. (2008), Ramachandran et al. (2013), Crockett et al. (2014)	
Autophagic vacuolar myopathy	5.15	AR		16p11.2	***CLN3*** *607042*	ceroid-lipofuscinosis, neuronal 3 (=battenin)	Cortese et al. (2014)	
Oculopharyngeal muscular dystrophy	5.16	AD	OPMD 164300	14q11.2-q13	***PABPN1*** *602279*	poly(A) binding protein, nuclear 1	Brais et al. (1995,1998) Robinson et al. (2005)	
Hereditary myopathy with early respiratory failure (Edström myopathy)	5.17	AD	HMERF 603689	2q24-3	***TTN*** *188840*	titin	Nicolao et al. (1999) Lange et al. (2005)	allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), CMH9 (group 10), CMD1G (group 10)
Epidermolysis bullosa simplex associated with late-onset muscular dystrophy	5.18	AR	MDEBS 226670	8q24-qter	***PLEC1*** *601282*	plectin	Gache et al. (1996) Smith et al. (1996) Wuyts et al. (1996)	allelic to LGMD2Q (group 1), myasthenic syndrome with plectin defect (group 11)
Muscle hypertrophy	5.19	AR	MSLHP 614160	2q32	***MSTN*** (=GDF8) *601788*	Myostatin (growth differentiation factor 8)	Schuelke et al. (2004)	
Fibrodysplasia ossificans progressiva	5.20	AD	FOP 135100	2q23-q24	***ACVR1*** *102576*	activin A receptor, type 1	Shore et al. (2006)	
HyperCKemia, idiopathic	5.21	AD	123320	3p25	***CAV3*** *601253*	caveolin-3	Carbone et al. (2000)	allelic to LGMD1C (group 1) and RMD2 (group 6), CMH (group 10)
X-linked myopathy with postural muscle atrophy	5.22	XR	XMPMA 300696	Xq26.3	***FHL1*** *300163*	four and a half LIM domain 1	Windpassinger et al. (2008)	allelic to Emery-Dreifuss MD X-linked type 2 (group 1) reducing body myopathy (group 3), XPMD (group 5)
Scapuloperoneal myopathy	5.23	XD	XPMD 300695	Xq26.3	***FHL1*** *300163*	four and a half LIM domain 1	Quinzil et al. (2008)	allelic to Emery-Dreifuss MD X-linked type 2 (group 1) reducing body myopathy (group 3), XMPMA (group 5)
Reducing body myopathy	5.24	XD	300717 300718	Xq26.3	***FHL1*** *300163*	four and a half LIM domain 1	Schessl et al. (2008), Shalaby et al. (2009)	allelic to EDMD6 (group 1), RSS (group 2), XPMA (group 5), XPMD (group 5)
Episodic muscle weakness, X-linked	5.25	XR	EMWX 300211	Xp22.3	*?*		Ryan et al. (1999)	
Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia	5.26	AD	IBMPFD 167320	9p13-p12	***VCP*** *601023*	valosin-containing protein	Watts et al. (2004), Haubenberger et al. (2005)	allelic to #1.42, IBMPFD (#4.17), ALS14 (#12.46) and CMT2Y (#14.63)
Myopathy with exercise intolerance, Swedish type	5.27	AR	HML 255125	12q24.1	***ICSU*** *611911*	iron-sulfur cluster scaffold homolog (E. coli)	Mochel et al. (2008)	
Late-onset axial myopathy related to RYR1	5.28	AD		19q13.1	***RYR1*** *180901*	Ryanodine receptor 1 (skeletal)	Løseth et al. (2013)	Allelic to items 3.12, 3.17, 3.18, 3.19, 3.20, 3.32
GROUP 6. MYOTONIC SYNDROMES								
Myotonic dystrophy (Steinert)	6.1	AD	DM1 160900	19q13	***DMPK*** *605377*	dystrophia myotonica-protein kinase	Renwick et al. (1971) Friedrich et al. (1987) Harley et al. (1992) Buxton et al. (1992) Aslanidis et al. (1992) Mahadevan et al. (1992) Fu et al. (1992) Brook et al. (1992)	
Myotonic dystrophy type 2 (proximal myotonic myopathy)	6.2	AD	DM2 (PROMM) 602668	3q21	***ZNF9*** *116955*	zinc finger protein 9	Ranum et al. (1998) Liquori et al. (2001)	

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Myotonia, dominant (Thomsen)	6.3	AD	*see under Ion channel muscle diseases*					
Myotonia, recessive (Becker)	6.4	AR	*see under Ion channel muscle diseases*					
Rippling muscle disease, dominant	6.5	AD	RMD1 600332	1q41	*?*		Stephan et al. (1994)	
Rippling muscle disease, dominant	6.6	AD	RMD2 606072	3p25	***CAV3*** *601253*	caveolin-3	Betz et al. (2001)	allelic to LGMD1C (group 1), hyper- Ckemia (group 5) , RMD2 (group 6), CMH (group 10)
Rippling muscle disease, recessive	6.7	AR	RMD2 606072	3p25	***CAV3*** *601253*	caveolin-3	Kubisch et al. (2003, 2005)	allelic to LGMD1C (group 1), hyper- Ckemia (group 5) , RMD2 (group 6), CMH (group 10)
Schwartz-Jampel syndrome	6.8	AR	SJS1 255800	1p34-p36.1	***HSPG*** *2142461 (perlecan)*	heparan sulfate proteoglycan 2 (perlecan)	Nicole et al. (1995, 2000)	
Brody disease	6.9	AR AD	601003	16p12	***ATP2A1*** *=SERCA1 108730*	ATPase, Ca++ transporting, cardiac muscle, fast twitch 1	Odermatt et al. (1996)	
Group 7. ION CHANNEL MUSCLE DISEASES								
A. Chloride channel								
Myotonia congenita, dominant (Thomsen)	7.1	AD	THD 160800	7q35	***CLCN1*** *118425*	muscle chloride channel	Koch et al. (1992b) George Jr et al. (1993)	allelic to Becker myotonia (group 7)
Myotonia, recessive (Becker)	7.2	AR	255700	7q35	***CLCN1*** *118425*	muscle chloride channel	Koch et al. (1992b)	allelic to Thomsen myotonia (group 7)
B. Sodium channel								
Hyperkalemic periodic paralysis	7.3	AD	hyperKPP 170500	17q23	***SCN4A*** *603967*	sodium channel, voltage-gated, type IV, alpha	Fontaine et al. (1990), Ptáček et al. (1991a) , Rojas et al. (1991), Miller et al. (2004)	allelic to HOKPP2 (group 7), PMC (group 7), K-aggravated myotonia (group 7)
Hypokalemic periodic Paralysis, type 2	7.4	AD	hypoKPP 170400	17q23	***SCN4A*** *603967*	sodium channel, voltage-gated, type IV, alpha	Bulman et al. (1999) Jurkat-Rott et al. (2000)	allelic to HYPP (group 7) , PMC (group 7), K-aggravated myotonia (group 7)
Paramyotonia congenita	7.5	AD	PMC 168300	17q23	***SCN4A*** *603967*	sodium channel, voltage-gated, type IV, alpha	Ptáček et al. (1991b, 1992a, 1993); Ebers et al. (1991) Koch et al. (1992a) Mc Clatchey et al. (1992)	allelic to HYPP (group 7) , HOKPP2 (group 7), K-aggravated myotonia (group 7)
Potassium-aggravated myotonia	7.6	AD	608390	17q23	***SCN4A*** *603967*	sodium channel, voltage-gated, type IV, alpha	Ptáček et al. (1992a, 1992b, 1994a) Heine et al. (1993) Lerche et al. (1993)	allelic to HYPP (group 7), HOKPP2 (group 7), PMC (group 7)
C. Calcium channel								
Hypokalemic periodic paralysis, type 1	7.7	AD	hypoKPP1 170400	1q31-q32	***CACNA1S*** *114208*	calcium channel, voltage-dependent, L type, alpha 1S subunit	Fontaine et al. (1994) Ptáček et al. (1994b) Jurkat-Rott et al. (1994) Elbaz et al. (1995)	
Acetazolamide responsive hereditary paroxysmal cerebellar ataxia (also listed in group 13 "Ataxias")	7.8	AD	APCA 108500	19p13	***CACNA1A*** *601011*	calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	von Brederlow et al. (1995) Vahedi et al. (1995)	allelic to EA2 (group 7), SCA6 (group 13)
Episodic ataxia type-2	7.9	AD	EA2 108500	19p13	***CACNA1A*** *601011*	calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	Ophoff et al. (1996) Jodice et al. (1997	allelic to APCA (group 7), SCA6 (group 13)
D. Potassium channel								
Hypokalemic periodic paralysis, type 3	7.10	AD	hypoKPP3 170400	11q13	***KCNE3*** *604433*	potassium voltage-gated channel, Isk-related family, member 3	Abbott et al. (2001)	

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Episodic ataxia/myokymia	7.11	AD	EA1 160120	12p13	***KCNA1*** *176260* *(voltage gated K + channel)*	potassium voltage-gated channel, shaker-related subfamily, member 1	Browne et al. (1994); Adelman et al. (1995)	
Thyrotoxic hypokalemic periodic paralysis	7.12		TTPP2 613239	17p11.2	***KCNJ18*** *613239*	Kir2.6 (inwardly rectifying potassium channel 2.6)	Ryan et al. (2010)	
Periodic paralysis, potassium sensitive cardiodysrythmic (Andersen syndrome)	7.13	*see LQ7 under hereditary cardiomyopathies (group 10/online only)*						
Long QT syndromes	7.14	*see under hereditary cardiomyopathies (group 10/online only))*						
GROUP 8. MALIGNANT HYPERTHERMIAS								
Malignant hyperthermia	8.1	AD	MHS1 145600	19q13.1	***RYR1*** 180901	ryanodine receptor 1 (skeletal)	MacLennan et al. (1990) McCarthy et al. (1990) Fujii et al. (1991) Gillard et al. (1991, 1992) Quane et al. (1993, 1994) Keating et al. (1994)	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), CNMDU1 (group 3)
Malignant hyperthermia	8.2	AD	MHS2 154275	17q11.2-q24	?		Levitt et al. (1992) Moslehi et al. (1998)	
Malignant hyperthermia	8.3	AD	MHS3 154276	7q21-q22	?		Iles et al. (1994)	
Malignant hyperthermia	8.4	AD	MHS4 600467	3q13.1	?		Sudbrak et al. (1995)	
Malignant hyperthermia	8.5	AD	MHS5 601887	1q31-q32	***CACNA1S*** (ex CACNL1A3) 114208	calcium channel, voltage-dependent, L type, alpha 1S subunit	Monnier et al. (1997)	allelic to HOKPP1 (group 7)
Malignant hyperthermia	8.6	AD	MHS6 601888	5p	?		Robinson et al. (1997)	
GROUP 9. METABOLIC MYOPATHIES								
A. Glycogen storage diseases								
Glycogen storage disease type II (Pompe disease) also listed in group 10 *Hereditary cardiomyopathies*	9.1	AR	GSD2 232300	17q25	***GAA*** *606800*	glucosidase, alpha; acid	Hers (1963); Martiniuk et al. (1990); Wokke et al. (1995)	allelic to GSDII (group 10)
Glycogen storage disease type IIIa	9.2	AR	GSD3 232400	1p21	***AGL*** *610860*	amylo-1, 6-glucosidase, 4-alpha-glucanotransferase (glycogen debranching enzyme)	Sheng et al. (1996)	
Glycogen storage disease type IV	9.3	AR	GSD4 232500	3p12	***GBE1*** *607839*	1,4-α-D = glucan 6-β-D-[1.4-D-glucano] transferase, branching enzyme 1 (glycogen branching enzyme) A	Brown et al. (1966) Bao et al. (1996) Bruno et al. (2004)	
Glycogen storage type V (McArdle)	9.4	AR	GSD5 232600	11q13	***PYGM*** *608455*	glycogen phosphorylase, muscle	Mommaerts et al. (1959) Schmidt et al. (1959) Lebo et al. (1984) Tsujino et al. (1993a)	
Glycogen storage type VII (Tarui)	9.5	AR	GSD7 232800	12q13	***PFKM*** *610681*	muscle-type phos-phofructokinase	Tarui et al. (1965) Nakajima et al. (1991), Howard et al. (1996)	
Glycogen storage disease type IXd (ex type VIII) or muscle phosphorylase kinase defiency	9.6	XR	GSD9D 300559	Xq13	***PHKA1*** *311870*	Phosphorylase b kinase, alpha subunit	Wehner et al. (1994) Burwinkel et al. (2004)	
Glycogenosis type XIV	9.7	AR	GSD14 612934	1p31	***PGM1*** *171900*	phosphoglucomutase 1	Stojkovic et al. (2009)	
Glycogenosis type XV	9.8	AR	GSD15 613507	3q24	***GYG1*** *603942*	Glycogenin 1	Moslemi et al. (2010)	Allelic to PGBM2 (group 9)

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Glycogen storage disease type 0	9.9	AR	GSD0b 611556	9q13	***GYS1*** *138570*	Glycogen synthase 1	Kolberg et al. (2007)	
Glycogen storage disease of heart, lethal congenital	9.10	AD	261740	7q36	***PRKAG2*** *602743*	protein kinase, AMP-activated (AMPK)	Burwinkel et al. (2005)	
Polyglucosan Body Myopathy 1 with or without immunodeficiency	9.11	AR	PGBM1 615895	20p13	***RBCK1*** *610924*	RanBP-type and C3HC4-type zinc finger containing 1 (heme-oxidized IRP2 ubiquitin ligase 1)	Nilsson et al. (2013)	
Polyglucosan Body Myopathy 2	9.12	AR	PGBM2	3q24	***GYG1*** *603942*	Glycogenin 1	Malfatti et al. (2014)	Allelic to GSD15 (group 9)
B. Glycolytic pathway								
Phosphoglycerate kinase deficiency	9.13	XR	300653	Xq13	***PGK1*** *311800*	phosphoglycerate kinase	DiMauro et al. (1981a, 1983) Rosa R (1982)	
Phosphoglycerate mutase deficiency	9.14	AR	GSD10 261670	7p12-p13	***PGAM2*** *612931*	phosphoglycerate mutase 2 (muscle)	DiMauro et al. (1981b) Edwards et al. (1989) Castella-Escola et al. (1990) Tsujino et al. (1993b)	
Lactate dehydrogenase-A deficiency	9.15	AR	GSD11 612933	11p15.4	***LDHA*** *150000*	lactate dehydrogenase A	Kanno et al. (1980) Scrable et al. (1990)	
Enolase deficiency	9.16	AD	GSD13 612932	17pter-p12	***ENO3*** *131370*	enolase 3, beta, muscle specific	Comi et al. (2001)	
C. Disorders of lipid metabolism								
Carnitine palmitoyl-transferase deficiency	9.17	AR	255110	1p32	***CPT2*** *600650*	carnitine palmitoyl transferase II	DiMauro et al. (1973) Finocchiaro et al. (1991) Taroni et al. (1993) Gellera et al. (1994)	
Primary systemic carnitine deficiency	9.18	AR	CDSP 212140	5q31	***SLC22A5*** *603377*	solute carrier family 22 , member 5	Nezu et al. (1999)	
Carnitine/acyl-carnitine translocase deficiency	9.19	AR	CACTD 212138	3p21.31	***SLC25A20*** *613698*	solute carrier family 25 (carnitine/acylcarnitine translocase), member	Huizing et al. (1997) Ogawa et al. (2000)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIA)	9.20	AR	GAIIA 231680	15q23-q25	***ETFA*** *608053*	electron-transfer-flavoprotein, alpha polypeptide	Indo et al. (1991) Freneaux et al. (1992)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIB)	9.21	AR	GAIIB 231680	19q13.3-q13.4	***ETFB*** *130410*	electron-transfer-flavoprotein, beta polypeptide	Colombo et al. (1994)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIC)	9.22	AR	GAIIC 231680	4q32-q35	***ETFDH*** *231675*	electron-transferring-flavoprotein dehydrogenase	Beard et al. (1993)	allelic to MADD (group 9)
Acyl-CoA dehydrogenase (very long chain) deficiency (VLCAD deficiency)	9.23	AR	ACADVLD 201475	17p13	***ACADVL*** *609575*	acyl-Coenzyme A dehydrogenase, very long chain	Aoyama (1993, 1995) Strauss et al. (1995) Mathur et al. 1999)	
ACAD9-deficient mild myopathy	9.24	AR	611126	3q21.3	***ACAD9*** *611103*	acyl-CoA dehydrogenase family member 9 **[M]**	Fragaki et al (2016)	
Triglyceride storage disease with ichthyosis [impaired long-chain fatty acid oxidation] (Chanarin-Dorfman syndrome)	9.25	AR	CDS 275630	3p25.3-p24.3	***ABHD5*** *(CGI-58)* *604780*	abhydrolase domain containing 5	Lefevre et al. (2001)	
Neutral lipid storage disease with myopathy without ichthyosis	9.26	AR	NLSDM 610717	11p15.5	***PNPLA2*** *609059*	adipose triglyceride lipase = desnutrin	Fischer et al. (2007)	
Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency (Lipid storage myopathy)	9.27	AR	MADD 231680	4q32-q35	***ETFDH*** *231675*	Electrontransferring-flavoprotein dehydrogenase	Olsen et al. (2007)	allelic to GAIIC (group 9)

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Recurrent myoglobinuria, autosomal recessive	9.28	AR	268200	2p25.1	***LPIN1*** *605518*	lipin 1 (phosphatidic acid phosphatase 1)	Zeharia et al. (2008)	
Infantile neuroaxonal dystrophy and neutral lipid storage disease with myopathy	9.29	AR	MMLA 251950	7q31.1	***PNPLA8*** *612123*	Patatin-like phospholipase domain containing 8 **[M]**	Saunders et al. (2015)	
Lipid storage myopathy due to Flavin Adenine Dinucleotide Synthetase Deficiency	9.30	AR	LSMFLAD 255100	1q21.3	***FLAD1*** *610595*	Flavin Adenine Dinucleotide Synthetase, S. cerevisiae, homolog of (M)	Taylor et al. (2014)	
GROUP 10/A HEREDITARY CARDIOMYOPATHIES non arrhythmogenic								
(a) Hypertrophic cardiomyopathies								
Familial hypertrophic cardiomyopathy, 1	10.1	AD	CMH1 192600	14q12	***MYH6*** *60710* ***MYH7*** *1 60760*	Myosin heavy chain 6 (alpha) or 7 (beta), cardiac muscle	Jarcho et al. (1989) Solomon et al. (1990) Tanigawa et al. (1990) Geisterfer-Lowrance et al. (1990)	allelic to CFTD (group 3), myosin storage myopathy (group 3), MPD1 (group 4), CMD1S (group 10)
Familial hypertrophic cardiomyopathy, 2	10.2	AD	CMH2 115195	1q32	***TNNT2*** *191045*	cardiac troponin T	Watkins et al. (1993) Thierfelder et al. (1994)	
Familial hypertrophic cardiomyopathy, 3	10.3	AD	CMH3 115196	15q22.1	***TPM1*** *191010*	tropomyosin-1	Thierfelder et al. (1994)	
Familial hypertrophic cardiomyopathy, 4	10.4	AD	CMH4 115197	11p11.2	***MYBPC3*** *600958*	cardiac myosin binding protein-C	Carrier et al. (1993) Bonne et al. (1995) Watkins et al. (1995)	allelic to congenital skeletal myopathy and fatal cardiomyopathy (group 3)
Familial hypertrophic cardiomyopathy, 6	10.5	AD	CMH6 600858	7q31	***PRKAG2*** *602743*	protein kinase, AMP-activated, gamma 2 non-catalytic subunit	Blair et al. (2001)	allelic to Glycogene storage disease of heart, lethal congenital (group 9)
Familial hypertrophic cardiomyopathy, 7	10.6	AD	CMH7	19q13.4	***TTNI3*** *191044*	cardiac troponin I	Kimura et al. (1997)	allelic to RCM1 and CMD2A (group 10)
Familial hypertrophic cardiomyopathy, 8	10.7	AD	CMH8 608751	3p21	***MYL3*** *160790*	myosin, light chain 3, alkali; ventricular, skeletal, slow	Poetter et al. (1996)	
Familial hypertrophic cardiomyopathy, 9	10.8	AD	CMH9 613765	2q24.3	***TTN*** *188840*	titin	Satoh et al. (1999)	allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), HMERF (group 5), CMD1G; (group 10)
Familial hypertrophic cardiomyopathy, 10	10.9	AD	CMH10 608758	12q23-q24	***MYL2*** *160781*	Myosin, light chain 2, regulatory, cardiac, slow	Poetter et al. (1996)	
Familial hypertrophic cardiomyopathy, 11	10.10	AD	CMH11 612098	15q14	***ACTC1*** *102540*	actin, alpha, cardiac muscle 1	Mogensen et al. (1999)	allelic to CMD1R (group 10)
Familial hypertrophic cardiomyopathy, 12	10.11	AD	CMH12 612124	11p15.1	***CSRP3*** *600824*	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Geier et al. (2008)	allelic to CMD1M (group 10)
Familial hypertrophic cardiomyopathy, 13	10.12	AD	CMH13 613243	3p21-p14	***TNNC1*** *191040*	Slow troponin C	Landstrom et al. (2008)	allelic to CMD1Z (group 10)
Familial hypertrophic cardiomyopathy, 14	10.13	AD	CMH14 613251	14q12	***MYH6*** *160710*	myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al. (2005)	allelic to CMD1EE (group 10)
Hypertrophic cardiomyopathy related to vinculin	10.14	AD	CMH15 613255	10q22	***VCL*** *193065*	vinculin (metavinculin)	Vasile et al. (2006)	allelic to CMD1U (group 10)
Hypertrophic cardiomyopathy	10.15	AD, digenic	CMH1 192600	20q13.3	***MYLK2*** *606566*	myosin light chain kinase 2	Davis et al. (2001)	
Hypertrophic cardiomyopathy	10.16	AD	CMH1 192600	3p25	***CAV3*** *601253*	caveolin-3	Hayashi et al. (2004) Fulizio et al. (2005)	allelic to LGMD1C (group 1), hyperCKemia (group 5) RMD2 (group 6)
Hypertrophic cardiomyopathy related to myozenin 2	10.17	AD	CMH16 613838	4q26	***MYOZ2*** *605602*	myozenin 2, or calsarcin 1, a Z disk protein	Osio et al. (2007)	

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Hypertrophic cardiomyopathy related to junctophilin	10.18	AD	CMH17 613873	20q13.12	***JPH2*** *605267*	junctophilin-2	Landstrom et al. (2007) Matsuhita et al. (2007)	
Hypertrophic cardiomyopathy related to phospholamban	10.19	AD	CMH18 613874	6q22	***PLN*** *172405*	phospholamban	Minamisawa et al. (2003) Landstrom et al. (2011)	Allelic to dilated cardiomyopathy (group 10)
Hypertrophic cardiomyopathy related calreticulin 3	10.20	AD	CMH19 61387	19p13.11	***CALR3*** *611414*	Calreticulin 3	Chiu et al. (2007)	
Hypertrophic cardiomyopathy related to nexilin	10.21	AD	CMH20 613876	1p31.1	***NEXN*** *613121*	Nexilin (F-actin binding protein)	Wang et al. (2010)	Allelic to CMD1CC (group 10)
Hypertrophic cardiomyopathy related to cardiac ankyrin repeat domain protein	10.22	AD		10q23.33	***ANKRD1*** *609599*	ankyrin repeat domain-protein 1 (cardiac)	Arimura et al. (2009)	Allelic to dilated cardiomyopathy (group 19)
Hypertrophic cardiomyopathy related to cardiac myopalladin	10.23	AD	CMH22 615248	10q21.1	***MYPN 608517***	Myopalladin	Purevjav et al. (2012)	allelic to CMD1KK (group 10A) and NEM11 (group 3)
Hypertrophic cardiomyopathy related to actinin-2	10.24	AD	CMH23 612158	1q43	***ACTN2*** *102573*	actinin alpha2	Chiu C et al. (2010)	Allelic to CMD1AA (group 10)
Hypertrophic cardiomyopathy related to ZASP	10.25	AD	CMH24 601493	10q23.2	***LDB3*** *605906*	LIM domain binding -3 (Z band alternatively spliced PDZ motif)	Theis et al. (2006)	Allelic to CMD1C (group 10A)
Hypertrophic cardiomyopathy related to TCAP	10.26	AD	CMH25 607487	17q12	***TCAP*** *604488*	Telethonin (titin-cap)	Hayashi et al. (2004)	Allelic to CMD1N (group 10A) and LGMD2G (group 1)
Mitochondrial hypertrophic cardiomyopathy related to *NDUFAF1*	10.27	AR		15q15.1	***NDUFAF1*** *606934*	NADH-ubiquinone oxidoreductase 1 alpha subcomplex **(M)**	Fassone et al. (2011)	
Mitochondrial hypertrophic cardiomyopathy related to *TSFM*	10.28	AR	COXPD3 610505	12q14.1	***TSFM*** *604723*	Ts translation elongation factor, mitochondrial **(M)**	Smeitink et al. (2006)	
Mitochondrial hypertrophic cardiomyopathy related to*AARS2*	10.29	AR	COXPD8 614096	6p21.1	***AARS2*** *612035*	Alanyl-tRNA synthetase 2, mitochondrial **(M)**	Götz et al. (2011)	
Mitochondrial hypertrophic cardiomyopathy related to *MRPL3*	10.30	AR	COXPD9 614582	3q21-q23	***MRPL3*** *607118*	Mitochondrial ribosomal protein L3 **(M)**	Galmiche et al. (2011)	
Mitochondrial hypertrophic cardiomyopathy, related to COX15	10.31	AR	CEMCOX2 615119	10q24	***COX15*** *603646*	COX15 homolog, cytochrome c oxidase assembly protein **(M)**	Antonicka et al. (2003)	
Mitochondrial hypertrophic cardiomyopathy related to *MTO1*	10.32	AR	COXPD10 614702	6q13	***MTO1*** *614667*	mitochondrial tRNA translation optimization 1 **(M)**	Ghezzi et al. (2012) Baruffini et al. (2013)	
Mitochondrial hypertrophic cardiomyopathy related to *MRPL44*	10.33	AR	COXPD16 615395	2q36.1	***MRPL44*** *611849*	Mitochondrial ribosomal protein L44 **(M)**	Carroll et al. (2013)	
(b) Dilated cardiomyopathies								
Dilated cardiomyopathy, 1A	10.34	AD	CMD1A 115200	1q21	***LMNA*** *150330*	lamin A/C	Fatkin et al. (1999)	allelic to EDMD2 (group 1), EDMD3 (group 1), LGMD1B (group 1), CMT2B1 (group 14), [+ FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, not in this table]
Dilated cardiomyopathy, 1B	10.35	AD	CMD1B 600884	9q13	*?*		Krajinovic et al. (1995)	
Dilated cardiomyopathy, 1C	10.36	AD	CMD1C 601493	10q22-q23	***LDB3 (ZASP)*** *605906*	LIM domain binding-3 (Z band alternatively spliced PDZ motif)	Bowles et al. (1996) Vatta et al. (2003) Arimura et al. (2004)	allelic to Markesbery-Griggs (group 4), MFM (group 5)
Dilated cardiomyopathy, 1D	10.37	AD	CMD1D 601494	1q32	***TNNT2*** *191045*	Troponin type 2 (cardiac)	Durand et al. (1995) Kamisago et al. (2000)	
Dilated cardiomyopathy, 1E with conduction disorder and arrhythmia	10.38	AD	CMD1E 601154	3p21	***SCN5A*** *600163*	Sodium channel, voltage-gated, type V, alpha	McNair et al. (2004)	allelic to LQT3 (group 10), Brugada syndrome (group 10), SSS1 (group 10)
Dilated cardiomyopathy, 1G	10.39	AD	CMD1G 604145	2q31	***TTN*** *188840*	Titin	Siu et al. (1999) Gerull et al. (2002) Itoh-Satoh et al. (2002)	allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), HMERF (group 5), CMH9 (group 10),
Dilated cardiomyopathy, 1H	10.40	AD	CMD1H 604288	2q14-q22	*?*		Jung et al. (1999)	

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Dilated cardiomyopathy, 1I	10.41	AD	CMD1I 604765	2q35	***DES*** *125660*	desmin	Li et al. (1999)	Allelic to LGMD1E (group 1), myofibrillar myopathy (group 5), ARVD7 (group 10/B)
Dilated cardiomyopathy, 1J	10.42	AD	CMD1J 605362	6q23-24	***EYA4*** *603550*	Eyes absent homolog	Schönberger et al. (2005a)	
Dilated cardiomyopathy, 1K	10.43	AD	CMD1K 605582	6q12-q16	*?*		Sylvius et al. (2001)	
Dilated cardiomyopathy, 1L	10.44	AD	CMD1L 606685	5q33	***SGCD*** *601411*	sarcoglycan, delta	Tsubata et al. (2000)	allelic to LGMD2F (group 1)
Dilated cardiomyopathy, 1M	10.45	AD	CMD1M 607482	11p15.1	***CSRP3*** *600824*	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Knoll et al. (2002)	allelic to CMH12 (group 10)
Dilated cardiomyopathy, 1N	10.46	AD	CMD1N 607487	17q12	***TCAP*** *604488*	Telethonin (titin-cap)	Knoll et al. (2002)	allelic to LGMD2G (group 1)
Dilated cardiomyopathy, 1O	10.47	AD	CMD1O 605569	12p12.1	***ABCC9*** *601439*	ATP-binding cassette, sub-family C (member 9)	Bienengraeber et al (2004)	
Dilated cardiomyopathy, 1P	10.48	AD	CMD1P 609909	6q22	***PLN*** *172405*	phospholamban	Schmitt et al. (2003) Haghighi et al. (2003, 2006)	allelic to hypertrophic cardiomyopathy CMH18 (group 10)
Dilated cardiomyopathy, 1Q	10.49	AD	CMD1Q 609915	7q22.3-q31.1	*?*		Schonberger et al. (2005b)	
Dilated cardiomyopathy, 1R	10.50	AD	CMD1R 613424	15q14	***ACTC1*** *102540*	actin, alpha, cardiac muscle 1	Olson et al. (1998) Mogensen et al. (1999)	allelic to CMH (group 10)
Dilated cardiomyopathy, 1S	10.51	AD	CMD1S 613426	14q12	***MYH7*** *160760*	myosin, heavy chain 7, cardiac muscle, beta	Kamisago et al. (2000)	allelic to allelic to CFTD (group 3), myosin storage myopathy (group 3), MPD1 (group 4), CMH1 (group 10)
Dilated cardiomyopathy, 1T	10.52	AD	CMD1T	12q22	***TMPO*** *188380*	Thymopoietin (lamina-associated polypeptide 2)	Taylor et al. (2005)	
Dilated cardiomyopathy, 1U	10.53	AD	CMD1U 613694	1q42.13	***PSEN2*** *600759*	Presenilin 2	Li et al. (2006)	
Dilated cardiomyopathy, 1W	10.54	AD	CMD1W 611407	10q22	***VCL*** *193065*	vinculin (metavinculin)	Olson et al. (2002) Vasile et al. (2006)	allelic to CMH 15 (group 10)
Dilated cardiomyopathy related to fukutin	10.55	AR	CMD1X 611615	9q31-q33	***FCMD*** *607440*	Fukutin	Murakami et al. (2006)	allelic to LGMD2L (group 1)
Dilated cardiomyopathy, 1Y	10.56	AD	CMD1Y 611878	15q22.2	***TPM1*** *191010*	Alpha-tropomyosin	Olson et al. (2010)	Allelic to CMH3 (group 10)
Dilated cardiomyopathy, 1Z	10.57	AD	CMD1Z 611879	3p21-p14	***TNNC1*** ***191040***	slow troponin C	Mogensen et al. (2004)	allelic to CMH13 (group 10)
Dilated cardiomyopathy, 1AA	10.58	AD	CMD1AA 612158	1q42-q43	***ACTN2*** *102573*	actinin alpha2	Mohapatra et al. (2003)	Allelic to CMH (group 10)
Dilated cardiomyopathy, related to DSG2	10.59	AD/AR	CMD1BB 612877	18q12.1	***DSG2*** *125671*	Desmoglein 2	Posch et al. (2008)	
Dilated cardiomyopathy related to nexilin	10.60	AD	CMD1CC 613122	1p32-p31	***NEXN*** *613121*	nexilin (<F-actin binding protein)	Hassel et al. (2009)	
Dilated cardiomyopathy related to ribonucleic acid binding protein	10.61	AD	CMD1DD 613172	10q25.3	***RBM20*** *613171*	RNA binding motif protein 20	Brauch et al. (2009)	
Dilated cardiomyopathy related to alpha-myosin heavy chain	10.62	AD	CMD1EE 613252	14q12	***MYH6*** *160710*	myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al. (2005)	Allelic to CMH1 (group 10)
Recessive neonatal isolated DC	10.63	AR	CMD1GG 613642	5p15.33	***SDHA*** *600857*	Succinate dehydrogenase complex, subunit a, flavoprotein **[M]**	Levitas et al. (2010)	
Dilated cardiomyopathy related to BAG3	10.64	AD	CMD1HH 613881	10q26.11	***BAG3*** ***603883***	BCL2-Associated Athanogene 3	Norton et al. (2011)	Allelic to MMF6 (group 5) and to CMT2 (group 14)
Dilated cardiomyopathy, related to alpha-crystallin	10.65	AD	CMD1II 615184	11q23.1	***CRYAB*** *123590*	Alpha B crystallin	Inagaki et al. (2006)	Allelic to myofibrillar myopathy (group 5)
Dilated cardiomyopathy related to laminin-alpha4	10.66	AD	CMD1JJ 615235	6q21	***LAMA4*** *600133*	laminin-alpha4	Knöll et al. (2007)	
Dilated cardiomyopathy related to myopalladin	10.67	AD	CMD1KK 615248	10q21.1	***MYPN*** *608517*	myopalladin	Duboscq-Bidot (2008)	
Dilated cardiomyopathy related to PRDM16	10.68	AD	CMD1LL LVNC8 615373	1p36.32	***PRDM1*** *605557*	PR Domain-Containing Protein 16	Arndt et al. (2013)	
Dilated cardiomyopathy related to *MYBPC3*	10.69	AD	CMD1MM 615396	11p11.2	***MYBPC3*** *600958*	Myosin-binding protein C	Hershberger et al. (2010)	Allelic to congenital myopathy and fatal cardiomyopathy (group 3), CMH4 (group 10)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Dilated cardiomyopathy related to RAF1	10.70	AD	CMD1NN 615916	3p25.2	***RAF1*** *164760*	V-Raf-1 Murine Leukemia Viral Oncogene Homolog 1	Dhandapany et al. (2014)	
Dilated cardiomyopathy related to integrin-linked kinase	10.71	AD		11p15.4	***ILK*** *602366*	integrin-linked kinase	Knöll et al. (2007)	
Dilated cardiomyopathy related to cardiac ankyrin repeat protein	10.72	AD		10q23.33	***ANKRD1*** *609599*	ankyrin repeat domain 1 (cardiac muscle)	Duboscq-Bidot (2009) Moulik et al. (2009)	
Dilated cardiomyopathy related to cardiac troponin I	10.73	AD	CMD2A 611880	19q34	***TNNI3*** *191044*	cardiac troponin I	Carballo et al. (2009)	allelic to CMH7 and RCM1
Dilated cardiomyopathy related to *GATAD1*	10.74	AR	CMD2B 614672	7q21.3	***GATAD1*** *614518*	GATA zinc finger domain containing 1	Theis et al. (2011)	
Dilated cardiomyopathy related to tafazzin	10.75	XR	CMD3A 300069	Xq28	***TAZ*** *300394*	tafazzin	Gedeon et al. (1995)	allelic to BTHS (group 10)
Dilated cardiomyopathy related to dystrophin	10.76	XR	CMD3B 302045	Xp21.2	***DMD*** *300377*	dystrophin	Muntoni et al. (1993) Milasin et al. (1996)	allelic to DMD (group 1)
Dilated cardiomyopathy related to nesprin-1	10.77	AD		6q25	***SYNE1*** *608441*	spectrin repeat containing, nuclear envelope 1 (nesprin-1	Puckelwartz et al. (2010)	Allelic to EDMD with nesprin-1 defect (group 1) SCAR8 (group 13), dilated APC with nesprin-1 defect (group 16)
Dilated cardiomyopathy related to *MURC*	10.78	AD		9q31.1	***MURC*** *(CAVIN4)*	Muscle-restricted colied-coil gene	Rodriguez et al. (2011)	
Dilated Cardiomyopathy related to *DOLK*	10.79	AR	CDGM1 610768	9q34.13	***DOLK*** *610746*	Dolichol kinase	Kranz et al. (2007) Lefeber et al. (2011	
(c) Restrictive cardiomyopathies								
Restrictive cardiomyopathy, 1	10.80	AD	RCM1 115210	19q34	***TTNI3*** *191044*	Cardiac troponin I	Mogensen et al. (2003)	allelic to CMH7 (group 10)
Restrictive cardiomyopathy, 2	10.81		RCM2 609578	10	*?*		Zhang et al. (2005)	
(d) Other non arrhythmogenic hereditary cardiomyopathies								
Pompe disease Glycogenosis, generalized, cardiac form (early and late onset) also listed in group 9	10.82	AR	GSDII 232300	17q25	***GAA*** *606800*	glucosidase, alpha; acid	Hers (1963) Martiniuk et al. (1990) Wokke et al. (1995)	allelic to GSDII (group 9)
Cardioskeletal myopathy with neutropenia and abnormal mitochondria (Barth syndrome)	10.83	XR	BTHS 302060	Xq28	***TAZ*** *300394*	tafazzin	Bolhuis et al. (1991) Bione et al. (1996)	allelic to CMD3A (group 10)
Left ventricular noncompaction	10.84	AD	LVNC 604169	18q12.1-q12.2	***DTNA*** *601239*	dystrobrevin, alpha	Ichida et al. (2001)	
Cardiovalvular dysplasia, X-linked (Myxomatous valvular dystrophy)	10.85	XR	XMVD 314400	Xq28	***FLNA*** *300017*	filamin A, alpha (actin binding protein 280)	Kyndt et al. (1998) Kyndt et al. (2007)	

10-B FOR ARRHYTHMOGENIC CARDIOMYOPATHIES AND RELATED SYNDROMES See online version of the gene table **http://www.musclegenetable.fr**

GROUP 11. CONGENITAL MYASTHENIC SYNDROMES

Slow channel syndromes	11.1	AD	CMS1A CMS1B 601462	2q24-q32	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1 muscle	Sine et al. (1995) Engel et al. (1996b) Croxen et al. (1997)	Allelic to other entries in group 11
	11.2	AD	CMS2A CMS2C 616313	17p11-p12	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1 muscle	Engel et al. (1996b) Gomez et al. (1996)	Allelic to other entries in group 11
	11.3	AD	CMS3A CMS3B CMS3C 616321	2q33-q34	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Gomez et al. (2002)	Allelic to other entries in group 11
	11.4	AD, AR	CMS4A CMS4B CMS4C 605809	17p13	***CHRNE 100725***	Cholinergic receptor, nicotinic, epsilon	Ohno et al. (1995) Gomez et al. (1995) Engel et al. (1996b) Croxen et al. (2002)	Allelic to other entries in group 11
Fast channel syndromes	11.5	AR	CMS1A CMS1B 608930	2q24-q32	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1 muscle	Wang et al. (1999) Shen et al. (2003)	Allelic to other entries in group 11
	11.6	AR	CMS3A, CMS3B, CMS3C 616322	2q33-q34	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Brownlow et al. (2001)	Allelic to other entries in group 11)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
	11.7	AR	CMS4A, CMS4B, CMS4C 616324	17p13	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Ohno et al. (1996)	Allelic to other entries in group 11
Acetylcholine receptor deficiency	11.8	AR	CMS2A CMS2C 616314	17p11-p12	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1 muscle	Quiram et al. (1999)	Allelic to other entries in group 11
	11.9	AR	CMS4A, CMS4B, CMS4C 616323	2q33-q34	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Shen et al. (2002)	Allelic to other entries in group 11
	11.10	AR	CMS3A, CMS3B, CMS3C 608931	17p13	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Engel et al. (1996a) Ohno et al. (1997)	Allelic to other entries in group 11
Congenital myasthenic syndrome related to end-plate acetylcholinesterase	11.11	AR	CMS5 603034	3p24.2	***COLQ*** *603033*	C-like tail subunit (single strand of homotrimer) of asymmetric acetylcholinesterase	Donger et al. (1998) Ohno et al. (1998, 1999, 2000)	
Congenital myasthenic syndrome related to choline acetyltransferase	11.12	AR	CMS6 254210	10q11.2	***CHAT*** *118490*	Choline acetyltransferase	Ohno et al. (2001) Maselli et al. (2003)	
Myasthenic syndrome, presynaptic, congenital, with or without motor neuropathy	11.13	AD	CMS7 616040	1q32.1	***SYT2*** *600104*	Synaptotagmin II	Herrmann et al. (2014)	
Familial limb girdle myasthenia related to agrin	11.14	AR	CMS8 615120	1p36.33	***AGRN*** *103320*	Agrin	Huzé et al. (2009)	
Congenital myasthenic syndromerelated to MuSK	11.15	AR	CMS9 616325	9q31-q32	***MUSK*** *601296*	Muscle-specific receptor tyrosine kinase	Chevessier et al. (2004)	
Familial limb girdle myasthenia related to *DOK7*	11.16	AR	CMS10 254300	4p16.2	***DOK7*** *610285*	Docking protein 7	Beeson et al. (2006); Selcen et al. (2008)	
Congenital myasthenic syndrome related to rapsyn	11.17	AR	CMS11 616326	11p11	***RAPSN*** *601592*	Rapsyn	Ohno et al. (2002) Ohno et al. (2003) Dunne et al. (2003)	
Familial limb girdle myasthenia with tubular aggregates related to *GFPT1*	11.18	AR	CMS12 610542	2p12-p15	***GFPT1*** *138292*	Glutamine-fructose-6-phosphate transaminase 1	Senderek et al. (2011)	
Familial limb-girdle myasthenia with tubular aggregates related to *DPAGT1*	11.19	AR	CMS13 614750	11q23.3	***DPAGT1*** ***191350***	Dolichyl-phosphate (UDP-N-acetylglucosamine) N-acetylglucosamine phosphotransferase 1 (GlcNAc-1-P transferase)	Belaya et al. (2012)	
Congenital myasthenic syndrome with tubular aggregates	11.20	AR	CMS14 616228	9q31.1	***ALG2*** *607905*	alpha-1,3/1,6-mannosyltransferase	Cossins et al (2013)	
Congenital myasthenic syndrome related to *ALG14*	11.21	AR	CMS15 607227	1p21.3	***ALG14*** *612866*	UDP-N-acetylglucosami-nyltransferase subunit	Cossins et al (2013)	
Myasthenic syndrome, acetazolamide-responsive	11.22	AR	CMS16 614198	17q23	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha	Tsujino et al. (2003)	Allelic to HOKPP2 (group 7), HYPP (group 7), PMC (group 7), K-aggravated myotonia (group 7)
Congenital myasthenic syndrome	11.23	AR	CMS17 616304	11p11.2	***LRP4*** *604270*	LDL receptor related protein 4	Ohkawara et al. (2014)	
Congenital myasthenic syndrome with intellectual disability and ataxia	11.24	*AD*	CMS18 616330	20p12.2	***SNAP25*** *600322*	synaptosome-associated protein 25	Shen et al. (2014)	
Congenital myasthenic syndrome type 19	11.25	AR	CMS19 616720	10q22.1	***COL13A1*** *120356*	collagen type XIII alpha 1 chain	Logan et al. (2015)	
Congenital myasthenic syndrome with episodic apnea	11.26	AR	CMS20 617143	2q12.3	***SLC5A7*** *608761*	solute carrier family 5 member 7	Bauche et al. (2016)	
Myasthenic Syndrome, Congenital, 21, Presynaptic	11.27	AR	CMS21 617239	10q11.23	***SLC18A3*** *600336*	Solute Carrier Family 18 (Vesicular Acetylcholine), Member 3	O'Grady et al. (2016)	

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Table (*continued*)

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Congenital myasthenic syndrome related to *PREPL* deficiency	11.28	AR	CMS22 616224	2p22.1	***PREPL*** *609557*	prolyl endopeptidase-like	Regal et al. (2014)	
Congenital myasthenic syndrome with nephrotic syndrome	11.29	AR	NPHS5 614199	3p21	***LAMB2*** *150325*	β2-laminin	Maselli et al. (2009)	
Escobar syndrome (multiple pterygium syndrome)	11.30	AR	EVMPS 265000	2q22-q44	***CHRNG*** *100730*	Cholinergic receptor, nicotinic, gamma	Hoffman et al. (2006) Morgan et al. (2006)	
Myasthenic syndrome, with plectin defect	11.31	AR		8q24-qter	***PLEC*** *601282*	Plectin	Banwell et al. (1999) Forrest et al. (2010) Selcen et al. (2011)	Allelic to LGMD2Q (group 1) MDEBS (group 5)
Congenital Myasthenia	11.32	AR		15q23	***MYO9A*** *604875*	myosin IXA	O'Connor et al. (2016)	
Congenital Myasthenic syndrome related to *GMPPB*	11.33	AR		3q21.3	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Belaya et al. (2015)	allelic to MEB, MDDGB14 (group 2), LGMD2T (group 1)
Presynaptic congenital myasthenic syndrome	11.34	AR		12p13.31	***VAMP1*** *185880*	Vesicle-Associated Membrane Protein 1	Shen et al. (2017) Salpietro et al. (2017)	allelic to SPAX1 (group 15)
Presynaptic congenital myasthenic syndrome	11.35	AR		20q13.33	***LAMA5*** *601033*	Laminin, Alpha 5	Maselli et al. (2017)	
GROUP 12. SPINAL MUSCULAR ATROPHIES MOTONEURON DISEASES								
Spinal muscular atrophy related to SMN1								
Spinal muscular atrophy , type I (Werdnig-Hoffman)	12.1	AR	SMA1 253300	5q11-q13	***SMN1*** *600354*	survival of motor neuron 1, telomeric	Gilliam et al. (1990) Melki et al. (1990a, 1994) Lefebvre et al. (1995) Bussaglia et al. (1995) Rodrigues et al. (1995) Roy et al. (1995) Hahnen et al. (1997)	allelic to SMA2 (group 12), SMA3 (group 12), SMA4 (group 12)
Spinal muscular atrophy, type II (intermediate)	12.2	AR	SMA2 253550	5q11-q13	***SMN1*** *600354*	survival of motor neuron 1, telomeric	Matthijs et al. (1996), Samilchuk (1996)	allelic to SMA1 (group 12), SMA3 (group 12), SMA4 (group 12)
Spinal muscular atrophy, type III (Kugelberg-Welander)	12.3	AR	SMA3 253400	5q11-q13	***SMN1*** *600354*	survival of motor neuron 1, telomeric	Brzustowicz et al. (1990) Melki et al. (1990b) Lefebvre et al. (1995)	allelic to SMA1 (group 12), SMA2 (group 12), SMA4 (group 12)
Spinal muscular atrophy, type IV, adult form	12.4	AR	SMA4 271150	5q11-q13	***SMN1*** *600354*	survival of motor neuron 1, telomeric	Brahe et al. (1995) Clermont et al. (1995)	allelic to SMA1 (group 12), SMA2 (group 12), SMA3 (group 12)
Distal spinal muscular atrophy, recessive								
Spinal muscular atrophy, distal autosomal recessive 1 (with respiratory distress)	12.5	AR	DSMA1 (SMARD1) 604320	11q13.2-q13.4	***IGHMBP2*** *600502*	immunoglobulin mu-binding protein 2	Grohmann et al. (1999, 2001)	
Spinal muscular atrophy, distal autosomal recessive 2	12.6	AR	DSMA2 605726	9p13.3	***SIGMAR1*** *601978*	sigma non-opioid intracellular receptor 1	Christodoulou et al. (2000) Li et al. (2015)	Allelic to ALS16 (group 12)
Spinal muscular atrophy, distal autosomal recessive 3	12.7	AR	DSMA3 607088	11q13	***?***		Viollet et al. (2004)	
Spinal muscular atrophy, distal autosomal recessive 4	12.8	AR	DSMA4 611067	1p36	***PLEKHG5*** *611101*	pleckstrin homology domain containing, family G (with RhoGef domain) member 5	Maystadt et al. (2006; 2007)	
Spinal muscular atrophy, distal related to *DNAJB2*	12.9	AR	DSMA5 614881	2q32-q34	***DNAJB2*** *(HSJ1)* *604139*	DnaJ (Hsp40) homolog, subfamily B, member 2	Blumen et al. (2012)	
Spinal muscular atrophy with congenital bone fractures 1	12.10	AR	SMABF1 *616866*	15q22.31	***TRIP4*** *604501*	thyroid hormone receptor interactor 4	Knierim et al. (2016)	MDCDC (group 2)
Spinal muscular atrophy with congenital bone fractures 2	12.11	AR	SMABF2 616867	10q22.1	***ASCC1*** *614215*	activating signal cointegrator 1 complex subunit 1	Knierim et al. (2016)	
Spinal muscular atrophy with progressive myoclonic epilepsy	12.12	AR	SMAPME 159950	8p22	***ASAH1*** *613468*	N-acylsphingosine amidohydrolase (acid ceramidase) 1	Zhou et al. (2012)	
Spinal muscular atrophy and cerebellar hypoplasia	12.13	AR		13q13.1	***EXOSC8*** *606019*	Exosome component 8	Boczonadi et al. (2014)	

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Distal spinal muscular atrophy, dominant								
Neuronopathy, distal hereditary motor, type I	12.14	AD	HMN1 182960	7q34-q36	***?***		Gopinath et al. (2007)	
Neuronopathy, distal hereditary motor, type I	12.15	AD	HMN2A 158590	12q24	***HSPB8*** *608014*	heat shock protein 8	Timmerman et al.; Irobi et al. (2004)	allelic to CMT2L (group 14)
Neuronopathy, distal hereditary motor, type II, adult juvenile	12.16	AD	HMN2B 608634	7q11.23	***HSPB1*** *602195*	heat-shock 27-kD protein-1	Evgrafov et al. (2004)	allelic to CMT2F (group 14)
Neuronopathy, distal hereditary motor, type IIC	12.17	AD	HMN2C 613376	5q11.2	***HSPB3*** *604624*	heat shock 27 kDa protein 3	Kolb et al. (2010)	
Distal Spinal Muscular Atrophy with Calf Predominance.	12.18	AD	HMN2D 615575	5q32	***FBXO38*** *608533*	F-box protein 38	Sumner et al. (2013)	
Distal spinal muscular atrophy, distal with upper limb predominance (type V)	12.19	AD	HMN5A (DSMAV) 600794	7p15	***GARS*** *600287*	glycyl tRNA synthetase	Christodoulou et al. (1995), Antonellis et al. (2003)	allelic to (CMT2D (group 14)
Distal spinal muscular atrophy type V	12.20	AD	HMN5A (DSMAV) 600794	11q13	***BSCL2*** *606158*	seipin	Windpassinger et al. (2004)	allelic to SPG17 (group 15)
Distal spinal muscular atrophy, type VB	12.21	AD	HMNS5B (DSMAVB) 614751	2p11.2	***REEP1*** *609139*	Receptor accessory protein 1	Beetz et al. (2012)	SPG 31 (group 15)
Dominant distal hereditary motor neuropathy	12.22	AD	dHMN	16q22.1	***AARS*** *601065*	alanyl-tRNA synthetase	Zhao et al. (2012)	Allelic to CMT2N (group 14)
Dominant distal hereditary motor neuropathy	12.23	AD	dHMN	14q32.2	***WARS*** *191050*	tryptophanyl-tRNA synthetase	Tsai et al. (2017)	
Spinal muscular atrophy, distal, with vocal cord paralysis (Harper-Young)	12.24	AD	HMN7A 158580	2q12.31	***SLC5A7 600****8761*	solute carrier family 5 (sodium/choline cotransporter), member 7	McEntagart et al. (2001) Barwick et al. (2012)	
Distal hereditary motor neuronopathy type VIIB	12.25	AD	HMN7B 607641	2p13	***DCTN1*** *601143*	dynactin 1	Puls et al. (2003)	Allelic to ALS related to DNCT1 (group 12)
Hereditary motor and sensory neuropathy V	12.26	AD	HMSN5 600361	4q34-q35	*?*		Muglia et al. (2008)	
Spinal muscular atrophy congenital non progressive of lower limbs	12.27	AD	SMAL 600175	12q24.1	***TRPV4*** *605427*	transient receptor potential cation channel, subfamily V, member 4	van der Vleuten et al. (1998) Auer-Grumbach et al. (2010) Deng et al. (2010)	allelic to SPSMA (group 10) allelic to CMT2C (group 14)
Spinal motor neuropathy	12.28			11q23.2	***RBM7*** *612413*	RNA binding motif protein 7	Giunta et al. (2016)	
Scapuloperoneal spinal muscular atrophy	12.29	AD	SPSMA 181405	12q.24	***TRPV4*** *605427*	transient receptor potential cation channel, subfamily V, member 4	Isozumi et al. (1996) Deng et al. (2010)	allelic to SMAL (group 10), CMT2C (group 14)
Spinal muscular atrophy, lower extremity, autosomal dominant	12.30	AD	SMALED 158600	14q32	***DYNC1H1*** *600112*	dynein, cytoplasmic 1, heavy chain 1	Harms et al. (2010, 2012)	CMT2O
Spinal muscular atrophy, lower extremity, autosomal dominant 2	12.31	AD	SMALED2 615290	9q22.31	***BICD2*** *609797*	bicaudal D homolog 2 (Drosophila)	Neveling et al. (2013) Oates et al. (2013) Peeters et al. (2013)	
Spinal muscular atrophy, late-onset, Finkel type	12.32	AD	SMAFK 182980	20q13	***VAP*** *605704*	vesicle-associated membrane protein-associated protein B and C	Nishimura et al. (2004a)	See also ALS8 (group 12)
Late-onset spinal motor neuronopathy, Jokela type	12.33	AD	SMAJ 615048	22q11.2-q13.2	***CHCHD10*** *(=C22orf16) 615903*	coiled-coil-helix-coiled-coil-helix domain containing 10 ***(M)***	Muller et al. (2014) Penttilä et al. (2012, 2015)	Allelic to FTDALS2 (group 12) and Mitochondrial myopathy (group 16)
Early onset spinal muscular atrophy lower extremity, autosomal dominant	12.34	AD		14 q	*?*		Harms et al. 2010	
Distal spinal muscular atrophy, X-linked								
Spinal muscular atrophy, distal, X-linked, related to *UBA1*	12.35	XR	SMAX2 301830	Xp11.23	***UBA1*** *(=UBE1) 314370*	ubiquitin-activating enzyme 1	Ramser et al. (2013) Diamini et al. (2013)	
Spinal muscular atrophy, distal, X-linked, related to *ATPTA*	12.36	XR	SMAX3 300489	Xq13-q21	***ATP7A*** *300011*	ATPase, Cu++ transporting, alpha polypeptide	Takata et al. (2004) Kennerson et al. (2010)	
Amyotrophic lateral sclerosis (ALS)								
Familial myotrophic lateral sclerosis 1 (dominant)	12.37	AD	ALS1 105400	21q22	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Siddique et al. (1991, 1996) Rosen et al. (1993)	
Familial amyotrophic lateral sclerosis 1 (recessive)	12.38	AR	ALS1 105400	21q22	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Andersen et al. (1995)	Allelic to IAHSP (group 15)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Amyotrophic lateral sclerosis 2 juvenile	12.39	AR	ALS2 205100	2q33	***ALS2*** *606352*	alsin	Hentati et al. (1994a) Yang et al. (2001) Hadano et al. (2001)	
Amyotrophic lateral sclerosis 3	12.40	AR	ALS3 606640	18q21	*?*		Hand et al. (2002)	
Amyotrophic lateral sclerosis 4	12.41	AD	ALS4 602433	9q34	***SETX*** *608465*	senataxin	Chance et al. (1998) Chen et al. (2004) Moreira et al. (2004)	Allelic to AOA2 (group 13)
Amyotrophic lateral sclerosis 5	12.42	AD	ALS5 602099	15q15-q21	*?*		Hentati et al. (1998)	
Amyotrophic lateral sclerosis 5	12.43	AR	ALS5 602099	5q21.1	***SPG11*** *610844*	Spatacsin	Orlacchio et al. (2010)	allelic to CMT2X (group 14), SPG11 (group 15)
Amyotrophic lateral sclerosis 6	12.44	AD	ALS6 608030	16p11.2	***FUS*** *137070*	fusion (involved in t(12;16) in malignant liposarcoma)	Sapp et al. (2003) Abalkhail et al. (2003), Kwiatkowski et al. (2009), Vance et al. (2009)	
Amyotrophic lateral sclerosis 7	12.45	AD	ALS7 608031	20p13	*?*		Sapp et al. (2003)	
Amyotrophic lateral sclerosis 8	12.46	AD	ALS8 608627	20q13	***VAPB*** *605704*	vesicle-associated membrane protein-associated protein B and C	Nishimura et al. (2004a, 2004b)	
Amyotrophic lateral sclerosis 9	12.47	AD	ALS9 611895	14q11.2	***ANG*** *105850*	angiogenin	Greenway et al. (2006) Wu et al. (2007)	
Amyotrophic lateral sclerosis 10, with or without frontotemporal dementia	12.48	AD	ALS10 612069	1p36.2	***TARDBP*** *605078*	TAR DNA-binding protein	Sreedharan et al. (2008)	
Amyotrophic lateral sclerosis 11,	12.49	AD	ALS11 612577	6q21	***FIG4 (KIAA0274)*** *609390*	Sac domain-containing inositol phosphatase 3	Chow et al. (2009)	Allelic to CMT4J (group 14)
Amyotrophic lateral sclerosis 12	12.50	AD AR	ALS12 613435	10p14	***OPTN*** *602432*	Optineurin	Maruyama et al. (2010)	
Amyotrophic lateral sclerosis 13	12.51	AD	ALS13 183090	12q24.12	***ATXN2*** *601517*	Ataxin 2	Elden et al. (2010) Daoud et al. (2011) Van Damme et al. (2011)	Allelic to spinocellular ataxia (group 13)
Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia	12.52	AD	ALS14 613954	9p13.3	***VCP*** *601023*	Valosin-containing protein	Johnson et al. (2011)	Allelic to #1.42 and IBMPFD (#4.17, #5.22)
Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	12.53	XD	ALS15 300857	Xp11.21	***UBQLN2*** *300264*	Ubiquilin 2	Deng et al. (2011)	
Amyotrophic lateral sclerosis 16, juvenile	12.54	AR	ALS16 614373	9p13.3	***SIGMAR1*** *601978*	sigma non-opioid intracellular receptor 1	Al-Saif et al. (2011)	Allelic to DSMA2 (group 12)
Amyotrophic lateral sclerosis 17	12.55	AD	ALS17 614696	3p11.2	***CHMP2B*** *609512*	charged multivesicular body protein 2B	Parkinson et al. (2006) Cox et al. (20010)	
Amyotrophic lateral sclerosis 18	12.56	AD	ALS18 614808	17p13.2	***PFN1*** *176610*	Profilin 1	Wu et al. (2012)	
Amyotrophic lateral sclerosis 19	12.57	AD	ALS19 615515	2q34	***ERBB4*** *600543*	V-ERB-B2 Avian Erythroblastic Leukemia Viral Oncogene Homolog 4	Takahashi et al. (2013)	
Amyotrophic lateral sclerosis 20	12.58	AD	ALS20 615426	12q13.13	***HNRPA1*** *164017*	heterogeneous nuclear ribonucleoprotein A1	Kim et al. (2013)	IBMPFD3 (group 3)
Familial amyotrophic lateral sclerosis.	12.59	AD	ALS21 606070	5q31	***MATR3*** *164015*	Matrin 3	Johnson et al. (2014)	Allelic to VCPDM (group 4)
Amyotrophic lateral sclerosis22	12.60	AD	ALS22 616208	2q35	***TUBA4A*** *191110*	Tubulin, Alpha-4A	Smith et al. (2014)	
Susceptibility to amyotrophic lateral sclerosis related to NEFH	12.61	AD	ALS1 105400	22q12.2	***NEFH*** *162230*	Neurofilament, heavy polypeptide	Al-Chalabie et al. (1999)	
Susceptibility to amyotrophic lateral sclerosis related to peripherin	12.62	AD	ALS1 105400	12q13.12	***PRPH*** *170710*	Peripherin	Gros-Louis et al. (2004), Leung et al. (2004)	
Susceptibility to amyotrophic lateral sclerosis related to dynactin 1	12.63	AD	ALS1 105400	2p13.1	***DCTN1*** *601143*	Dynactin 1	Munch et al. (2005)	Allelic to HMN7B (group 12)

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Table (*continued*)

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Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.64	AD	FTDALS1 (ALSFTD) 105550	9p21.2	***C9orf72*** *614260*	chromosome 9 open reading frame 72	Morita et al. (2006) DeJesus-Hernandez (2011)	
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.65	AD	FTDALS2 615911	22q11	***CHCHD10*** *(=C22orf16)* *615903*	Coiled-coil-helix -coiled-coil-helix domain containing 10 ***(M)***	Bannwarth et al. (2014)	Allelic to mitochondrial myopathy (group 16)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.66	AD	FTDALS3 66437	5q35.3	***SQSTM1*** *601539*	Sequestosome 1	Fecto et al. (2011)	
Others								
Kennedy disease	12.67	XR	SBMA 313200	Xq13	***AR*** *313700*	androgen receptor	Fishbeck et al. (1986) La Spada et al. (1991)	
Lethal Congenital Contracture Syndrome 1	12.68	AR	LCCS1 253310	9q34	***GLE1*** *603371*	GLE1 RNA export mediator homolog (yeast)	Makela-Bengs et al. (1998) Nousiainen et al. (2008)	
Lethal Congenital Contracture Syndrome 2	12.69	AR	LCCS2 607598	12q13	***ERBB3*** *190151*	v-erb-b2 erythroblastic leukemia viral oncogene homolog 3 (avian)	Narkis et al. (2007)	
Lethal Congenital Contracture Syndrome 3	12.70	AR	LCCS3 611359	19p13	***PIP5K1C*** *606102*	phosphatidylinositol-4-phosphate 5-kinase, type I, gamma	Narkis et al. (2007)	
Spinal muscular atrophy with pontocerebellar hypoplasia, type 1b	12.71	AR	PCH1B 614678	9p13.2	***EXOSC3*** *606489*	exosome component 3	Rudnik-Schöneborn et al. (2013)	
Spinal muscular atrophy with or without pontocerebellar hypoplasia	12.72	AR	PCH1 607596	14q32	***VRK1*** *602168*	Vaccinia-related kinase 1	Renbaum et al. (2009) Stoll et al. (2016)	
Pontocerebellar hypoplasia with spinal muscular atrophy	12.73	AR	HSMN6B 616505	5q22.1	***SLC25A46*** *610826*	Solute Carrier Family 25, Member 46	van Dijk et al. (2017)	
Brown-Vialetto-van Laere syndrome 1	12.74	AR	BVVLS1 211530	20p13	***SLC52A3*** *613350*	Solute carrier family 52, riboflavin transporter, member 3	Green et al. (2010) Bosch et al. (2011) Johnson et al. (2010)	
Brown-Vialetto-van Laere syndrome 2	12.75	AR	BVVLS2 614707	8q24	***SLC52A2*** *607882*	Solute carrier family 52, riboflavin transporter, member 2	Green et al. (2010) Bosch et al. (2011) Johnson et al. (2012)	
Late onset spinal muscular atrophy related to *HEXB*	12.76	AD		5q13.3	***HEXB*** *606873*	hexosaminidase B	Rattay et al. (2013)	Sandhoff disease, adult type (OMIM #268800)

GROUP 13. HEREDITARY ATAXIAS See online version of the gene table at http://musclegenetable.fr

GROUP 14. HEREDITARY MOTOR SENSORY NEUROPATHIES (HMSN)

A. Charcot-Marie-Tooth neuropathy, type I (demyelinating)

Autosomal dominant (AD-CMT1)

Type 1A	14.1	AD	CMT1A 118220	17p11.2	***PMP22*** *601097*	peripheral myelin protein 22	Vance et al. (1989) Patel et al. (1992) Matsunami et al. (1992); Timmerman et al. (1990, 1992); Valentijn et al. (1992) Roa et al. (1993a)	allelic to CMT1E (group14), HNPP (group14), DSS (group14)
Type 1B	14.2	AD	CMT1B 118200	1q22	***MPZ*** *159440*	myelin protein zero	Bird et al. (1982) Guiloff et al. (1982) Hayasaka et al. (1993a) Kulkens et al. (1993)	allelic to CMT2I (group14), CMT2J (group14), DSS (group14), CMT4E (group14)
Type 1C	14.3	AD	CMT1C 601098	16p13	***LITAF*** *603795*	lipopolysaccharide-induced TNF factor	Street et al. (2002, 2003)	
Type 1D	14.4	AD	CMT1D 607678	10q21.1	***EGR2*** *129010*	early growth response 2 (Krox-20 homolog)	Warner et al. (1998), Street et al. (2003)	allelic to CMT4E (group 14), DSS (group 14)
Type 1E (with deafness)	14.5	AD	CMT1E 118300	17p11.2	***PMP22*** *601097*	peripheral myelin protein 22	Kovach et al. (1999); Boerkoel et al. (2002)	allelic to CMT1A (group 14), DSS (group14)
Hereditary Neuropathy with Liability to Pressure Palsies	14.6	AD	HNPP 162500	17p11.2	***PMP22*** *601097*	peripheral myelin protein P22	Chance et al. (1993) Nicholson et al. (1994) Mariman et al. (1994)	allelic to CMT1A (group 14) CMT1E (group 14), HNPP (group 14), DSS (group 14)

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Type 1F	14.7	AD	CMT1F 607734	8p21	***NEFL*** *162280*	neurofilament, light polypeptide 68 kDa	Jordanova et al. (2003)	allelic to CMT2E (group14)
CMT with congenital vertical talus	14.8	AD	CVT 192950	2q31-q32	***HOXD10*** *(HOX4)* *142984*	homeobox D10	Shrimpton et al. (2004)	
Slowed nerve conduction velocity	14.9	AD	NCV 608236	8p23	***ARHGEF10*** *608136*	Rho guanine-nucleotide exchange factor-10	De Jonghe, et al. (1999) Verhoeven, et al. (2003)	
Neuropathy, hereditary, with or without age-related macular degeneration	14.10	AD	HNARMD 608895	14q32.12	***FBLN5*** *604580*	Fibulin 5	Auer-Grumbach et al. (2011)	
CMT1 related to *PMP2*	14.11	AD	CMTDI	8q21.13	***PMP2*** ***170715***	Peripheral Myelin Protein 2	Hong et al. (2016)	
Dominant intermediate (CMTDI)								
Type A	14.12	AD	CMTDIA 606483	10q24.1-q25.1	***?***		Verhoeven et al. (2001)	
Type B	14.13	AD	CMTDIB 606482	19p12-13.2	***DNM2*** *602378*	dynamin 2	Zuchner et al. (2005)	allelic to CNM (group 3) and CM (group 2)
Type C	14.14	AD	CMTDIC 608323	1p35	***YARS*** *603623*	tyrosyl-tRNA synthetase	Jordanova et al. (2003, 2006)	
Type D	14.15	AD	CMTDID 607791	1q22	***MPZ*** *159440*	myelin protein zero	Mastaglia et al. (1999)	allelic to CMT1B, CMT4E, CMT2I, CMT2J, DSS (group 14)
Type E with glomerulopathy	14.16	AD	CMTDIE 614455	14q32-33	***INF2*** *610982*	Inverted formin 2	Boyer et al. (2011)	
Type F	14.17	AD	CMTDIF 615185	3q28-q29	***GNB4*** *610863*	guanine nucleotide-binding protein, beta-4	Soong et al. (2013)	
Autosomal recessive (AR-CMT1 or CMT4)								
CMT, type 4A	14.18	AR	CMT4A (=CMT2H) 214400	8q13-q21	***GDAP1*** *606598*	ganglioside induced differentiation associated protein1 (connexin 32)	Ben Othmane et al. (1993b) Baxter et al. (2002) Cuesta et al. (2002) Nelis et al. (2002)	allelic to CMT2K and Autosomal recessive CMT2C (group 14)
CMT, type 4B1	14.19	AR	CMT4B1 601382	11q22	***MTMR2*** *603557*	myotubularin-related protein-2	Bolino et al. (1996, 2000) Previtali et al. (2003)	
CMT, type 4B2	14.20	AR	CMT4B2 604563	11p15	**SBF2** (=MTMR13) 607697	SET binding factor 2	Azzedine et al (2003); Senderek et al. (2004)	
CMT, type 4B3	14.21	AR	CMT4B3 615284	22q13.33	**SBF1** (=MTMR5) 603560	SET binding factor 1	Nakhro et al. (2013)	
CMT, type 4C	14.22	AR	CMT4C 601596	5q32	***SH3TC2*** *608206* *(ex-KIAA1985)*	SH3 domain and tetratricopeptide repeats 2	LeGuern et al. (1996), Senderek et al. (2003)	
CMT, type 4D	14.23	AR	CMT4D (HMNSL) 601455	8q24	***NDRG1*** *605262*	Nmyc downstream regulated gene 1	Kalaydjieva et al. (1996, 2000) Hunter et al. (2003)	Allelic to HMNSL (group 14)
CMT, type 4E (congenital hypomyelinating myopathy)	14.24	AR	CMT4E 605253	10q21.1	***EGR2*** *129010*	early growth response 2 (Krox-20 homolog)	Warner et al. (1998)	allelic to CMT1D (group 14)
CMT, type 4E (congenital hypomyelinating myopathy)	14.25	AR	CMT4E 605253	1q22	***MPZ*** *159440*	myelin protein zero	Warner et al. (1996)	allelic to CMT1B (group 14), CMT2I (group 14), CMT2J (group 14), DSS (group 14)
CMT, type 4F	14.26	AR	CMT4F 145900	19q13	***PRX*** *605725*	periaxin	Delague et al. (2000) Guilbot et al. (2001)	allelic to DSSE (group14)
CMT, type 4G (type Russe)	14.27	AR	CMT4G 605285	10q22	***HK1*** *142600*	Hexokinase 1	Rogers et al. (2000) Thomas et al. (2001) Hantke et al. (2009) Sevilla et al. (2013)	
CMT, type 4H	14.28	AR	CMT4H 609311	12p11.21	***FGD4*** *611104*	frabin	De Sandre-Giovannoli et al. (2005) Delague et al. (2007) Stendel et al. (2007)	
CMT, type 4J	14.29	AR	CMT4J 611228	6q21	**FIG4** (=KIAA0274) 609390	Sac domain-containing inositol phosphatase 3	Chow et al. (2007)	Allelic to ALS 11 (group 12)
CMT, type 4K	14.30	AR	CMT4K	9q34.2	***SURF1*** *185620*	surfeit 1 **[M]**	Echaniz-Laguna et al. (2013)	

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
X-linked CMT1								
CMT1, X-linked 1	14.31	XD	CMTX1 302800	Xq13	***GJB1*** *304040*	gap junction protein, beta 1, 32 kDa (connexin 32)	Bergoffen et al. (1993) Bone et al. (1995)	allelic to DSS (group 14)
CMT1, X-linked 2	14.32	XR	CMTX2 302801	Xp22.2	***?***		Ionasecu et al. (1992)	
CMT1, X-linked 3	14.33	XR	CMTX3 302802	Xq26	***78 kb Chro8 insertion***	78 kb inter-chromosomal insertion (from chro 8q24.3)	Ionasecu et al. (1992) Huttner et al. (2006) Brewer et al. (2016)	
CMT1, X-linked 4 (Cowchock syndrome)	14.34	XR	CMTX4 310490	Xq24-q26	***AIFM1*** *300169*	Apoptosis-inducing factor, mitochondrion-associated, 1 **[M]**	Priest et al. (1995) Rinaldi et al. (2012)	
CMT1, X-linked 5 (with hearing loss and optic neuropathy)	14.35	XR	CMTX5 311070	Xq22-q24	***PRPS1*** *311850*	phosphoribosyl pyrophosphate synthetase 1	Kim et al (2007)	
CMT1, X-linked 6	14.36	XD	CMTX6 300905	Xp22.11	***PDK3*** *300906*	Pyruvate dehydrogenase kinase, isoenzyme 3	Kennerson et al. (2013)	
Dejerine-Sottas syndrome (DSS or CMT3)								
Dejerine-Sottas hypertrophic neuropathy, dominant	14.37	AD	DSSA 145900	17p11.2	***PMP22*** *601097*	peripheral myelin protein 22	Roa et al. (1993b)	allelic to CMT1A (group 14) CMT1E (group 14), HNPP (group 14)
Dejerine-Sottas hypertrophic neuropathy, dominant	14.38	AD	DSSB 145900	1q21-q23	***MPZ*** *159440*	myelin protein zero	Hayasaka et al. (1993b)	allelic to CMT1B (group 14), CMT2I (group 14), CMT2J (group 14), CMT4E (group 14)
Dejerine-Sottas hypertrophic neuropathy, dominant	14.39	AD (digenic)	DSSC 145900	10q21. Xq13	***EGR2*** *129010* and ***GJB1*** *304040*	early growth response 2 (Krox-20 homolog) and gap junction protein, beta 1, 32k Da (connexin 32)	Chung et al. (2005)	allelic to CMTX1 (group 14)
Dejerine-Sottas hypertrophic neuropathy, recessive	14.40	AR	DSSE (=CMT4F) 145900	19q13	***PRX*** *605725*	periaxin	Delague et al. (2000); Boerkoel et al. (2001)	allelic to CMT4F (group 14)
B. Charcot-Marie-Tooth neuropathy, type 2 (axonal) = CMT2 autosomal dominant								
Type 2A1	14.41	AD	CMT2A1 118210	1p36.2	***KIF1B*** *605995*	kinesin family member 1B	Zhao, C. et al. (2001a)	
Type 2A2	14.42	AD	CMT2A2A 609260	1p36.2	***MFN2*** *608507*	mitofusin 2	Ben Othmane et al. (1993a) Züchner, et al. (2004)	
Type 2B	14.43	AD	CMT2B 600882	3q21	***RAB7*** *602298*	RAB7, member of RAS oncogene family)	Kwon et al. (1995) Pericak-Vance et al. (1997) Kok et al. (2003)	
Type 2C	14.44	AD	CMT2C 606071	12q23-q24	***TRPV4*** *600175*	transient receptor potential cation channel, subfamily V, member 4	Klein et al. (2003) McEntagart et al. (2005) Auer-Grumbach et al. (2010) Deng et al. (2010) Landoure et al. (2010)	
Type 2D	14.45	AD	CMT2D 601472	7p15	***GARS*** *600287*	glycyl tRNA synthetase	Ionasescu et al. (1996) Antonellis et al. (2003)	allelic to DSMAV (group12)
Type 2E	14.46	AD	CMT2E 607684	8p21	***NEFL*** *162280*	neurofilament, light polypeptide 68 kDa	Birouk et al. (2003) Claramunt et al. (2005)	allelic to SMAL and SPSMA (group 12)
Type 2F	14.47	AD	CMT2F 606595	7q11-q21	***HSPB1*** *602195*	heat-shock 27-kD protein-1	Ismailov et al. (2001) Evgrafov et al. (2004)	
Type 2G	14.48	AD	CMT2G 608591	12q12-q13	*?*	?	Nelis et al. (2004)	
Type 2H	14.49	AD	CMT2H 607731	8q21.3	*?*	?	Barhoumi et al. (2001)	maybe allelic to CMT4A (group14)
Type 2I (late onset)	14.50	AD	CMT2I 607677	1q22	***MPZ*** *159440*	myelin protein zero	Auer-Grumbach et al. (2003)	allelic to CMT1B (group 1), CMT2J (group 14), DSS (group 14), CMT4E (group 14)
Type 2J (with hearing loss and pupillary abnormality)	14.51	AD	CMT2J 607736	1q22	***MPZ*** *159440*	myelin protein zero	De Jonghe et al. (1999) Chapon et al. (1999)	allelic to CMT1B (group 14), CMT2J (group 14), DSS (group 14), CMT4E (group 14)
Type 2K	14.52	AD, AR	CMT2K 607831	8q13-q21	***GDAP1*** *606598*	ganglioside-induced differentiation-associated protein 1	Nelis et al. (2002) Birouk et al. (2003) Claramunt et al. (2005)	allelic to CMT4A and ARCMT2C (group 14)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Type 2L	14.53	AD	CMT2L 608673	12q24	***HSPB8*** *608014*	heat shock protein 8	Tang et al. (2004, 2005)	allelic to HMN2A (group 12)
Type 2N	14.54	AD	CMT2N 613287	16q22.1	***AARS*** *601065*	AARS alanyl-tRNA synthetase	Latour et al. (2010)	
Type 2O	14.55	AD	CMT2O 614228	14q32.31	***DYNC1H1*** *600112*	dynein, cytoplasmic 1, heavy chain 1	Weedon et al. (2011)	
Type 2P	14.56	AD	CMT2P 614436	9q33.3	***LRSAM1*** *610933*	leucine rich repeat and sterile alpha motif containing 1	Guernsey et al. (2010) Weterman et al. (2012) Nicolaou et al. (2012)	
Type 2Q	14.57	AD	CMT2Q 615025	10p14	***DHTKD1*** *614984*	dehydrogenase E1 and transketolase domain containing 1	Xu et al. (2012)	
Type 2U	14.58	AD	CMT2U 616280	12q13.3	***MARS*** *156560*	methionyl-tRNA synthetase	Gonzalez et al. (2013)	
Type 2V	14.59	AD	CMT2V 616491	17q21.2	***NAGLU*** *609701*	N-acetyl-alpha-glucosaminidase	Tetreault et al. (2015)	
Type 2W	14.60	AD	CMT2W 616625	5q31.3	***HARS*** *142810*	histidyl-tRNA synthetase	Vester et al. (2013) Safka-Brozkova (2015)	
Type 2Y	14.61	AD	CMT2Y 616687	9p13.3	***VCP*** *601023*	Valosin-containing protein	Gonsalez et al. (2014)	Allelic to 1.48, IBMPFD (groups 4 and 5), ALS14 (group 12)
Type 2Z	14.62	AD	CMT2Z 616688	2q12.2	***MORC2*** ***616661***	MORC family CW-type zinc finger 2	Albulym et al. (2015) Sevilla et al. (2016)	
Hereditary motor and sensory neuropathy, Okinawa type	14.63	AD	HMSN0/ HMNSP 604484	3q13	***TFG*** *602498*	TRK-fused gene	Takeshima et al. (1997, 1999) Maeda et al. (2007) Ishiura et al. (2012)	
CMT2 related to *KIF5A*	14.64	AD		12q13.13	***KIF5A*** *602821*	kinesin family member 5A	Liu et al. (2014)	Allelic to SPG10 (group 15)
Early onset axonal neuropathy with sensory ataxia	14.65	AD	CMT2	1q13.3	***DGAT2*** 606983	diacylglycerol O-acyltransferase 2	Hong et al. (2016)	
Charcot-Marie-Tooth disease, axonal, type 2CC	14.66	AD	CMT2CC 616924	22q11.2	***NEFH*** *162230*	Neurofilament Protein, Heavy Polypeptide	Rebelo et al. (2016)	
Charcot-Marie-Tooth disease, axonal, related to BAG3	14.67	AD		10q25-q26	***BAG3*** *603883*	BCL2-associated athanogene 3	Noury et al. (2017)	Allelic to MFM6 (group 5) and to CMH1HH (group 10A)
CMT2 Autosomal recessive								
AR-CMT2A	14.68	AR	CMT2B1 605588	1q21.2	***LMNA*** *150330*	lamin A/C	Bouhouch et al. (1999) De Sandre et al. (2002) Worman and Bonne (2007)	allelic to EDMD2 (group 1), EDMD3 (group 1), LGMD1B (group 1), [+ FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, not in this table]
AR-CMT2B	14.69	AR	CMT2B2 605589	19q13	***MED25*** *610697*	mediator complex subunit 25	Leal et al. (2001, 2009)	allelic to CMT4A, and CMT2K (group 14)
AR-CMT2 related to *DNAJB2*	14.70	AR		2q35	***DNAJB2 (=HSJ1)*** *604139*	DnaJ (Hsp40) homolog, subfamily B, member 2	Gesse et al. (2014)	Allelic to SMA (group 12)
Axonal neuropathy with neuromyotonia	14.71	AR	NMAN 137200	5q23.3	***HINT1*** *601314*	histidine triad nucleotide binding protein 1	Zimon et al. (2012)	
Axonal neuropathy intermediate recessive B	14.72	AR	CMTRIB 613641	16q23.1	***KARS*** *601421*	Lysyl-tRNA synthetase	McLaughlin et al. (2010)	
Axonal neuropathy intermediate recessive C	14.73	AR	CMTRIC 615376	1p36	***PLEKHG5*** *611101*	Pleckstrin homology domain containing, family G (with RhoGef domain) member 5	Azzedine et al. (2013) Kim et al. (2013)	Allelic to DSMA4 (group 12)
CMT recessive intermediate D	14.74	AR	CMTRID 616039	12q24.31	***COX6A1*** *602072*	cytochrome c oxidase subunit VIa polypeptide 1 **[M]**	Tamiya et al. (2014)	
Type 2R	14.75	AR	CMT2R 615490	4q31.3	***TRIM2*** *614141*	tripartite motif containing 2	Ylikallio et al. (2013) Pehlivan et al. (2015)	
Autosomal recessive CMT axonal type 2S	14.76	AR	CMT2S 616155	11q13.3	***IGHMBP2*** *600502*	immunoglobulin mu binding protein 2	Cottenie et al. (2014) Schottmann et al. (2015)	Allelic to DSMA1 (group 12)
Type 2T	14.77	AR	CMT2T 617017	3q25.2	***MME*** *120520*	membrane metalloendopeptidase	Higuchi et al. (2016)	

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Type 2X	14.78	AR	CMT2X 616668	15q21.1	***SPG11*** *610844*	Spatacsin	Montecchiani et al. (2015)	ALS5 (group 12), SPG11 (group 15)
Early-onset axonal Charcot-Marie-Tooth with ataxia	14.79	AR	AOA4 616267	19q13.33	***PNKP*** 605610	polynucleotide kinase 3′-phosphatase	Pedroso et al. (2015)	
Charcot-Marie-Tooth disease, axonal	14.80	AR		10q22.1	***SGPL1*** *603729*	Sphingosine-1-Phosphate Lyase 1	Atkinson et al. (2017)	allelic to Nephrotic Syndrome 14 (# 617575)
C. CMT Distal = Distal hereditary motor neuropathies (dHMN) = spinal CMT or distal spinal muscular atrophy (DSMA) See under MOTOR NEURONE DISEASES (Group 12)								
D. Other HSMN syndromes								
Hereditary sensory and autonomic neuropathy type I	14.81	AD	HSAN1 162400	9q22.1-q22.3	***SPTLC1*** *605712*	serine palmitoyltransferase long chain base subunit 1	Nicholson et al. (1996) Bejaoui et al. (2001) Dawkins et al. (2001)	
Hereditary sensory and autonomic neuropathy type IB with cough and gastroesophageal reflux	14.82	AD	HSAN1B 608088	3p24-p22	***?***		Kok et al. (2004)	
Hereditary sensory and autonomic neuropathy type I	14.83	AD	HSN IC 613640	14q24.3	***SPTLC2*** *605713*	serine palmitoyltransferase long chain base subunit 2	Rotthier et al. (2010)	
Hereditary sensory neuropathy type I.D	14.84	AD	HSN ID 613708	14q22.1	***ATL1*** *606439*	atllastin GTPase 1	Guelly et al. (2011)	Allelic to SPG3A (group 15)
Neuropathy, hereditary sensory, type IF	14.85	AD	HSN IF 615632	11q13.1	***ATL3*** 609369	atllastin GTPase 3	Kornak et al. (2014)	
Hereditary sensory and autonomic neuropathy type II	14.86	AR	HSAN2 201300	12p.13	***WNK1*** *605232*	WNK lysine deficient protein kinase 1	Lafreniere et al. (2004) Shekarabi et al. (2008)	
Hereditary sensory and autonomic neuropath type IIB	14.87	AR	HSAN2B 613115	5p15.1	***FAM134B*** 613114	family with sequence similarity 134 member B	Kurth et al. (2009)	
Hereditary sensory neuropathy, type IIC	14.88	AD	HSN IIC 614213	2q37.3	***KIF1A*** *601255*	kinesin family member 1A	Riviere et al. (2011)	Allelic to SPG30 (group 15)
Neuropathy, hereditary sensory and autonomic, type IId,	14.89	AR	HSAN2D 24300	2q24.3	***SCN9A*** 603415	sodium voltage-gated channel alpha subunit 9	Yuan et al. (2013)	
Hereditary sensory and autonomic neuropathy type III (Familial dysautonomia, Riley-Day syndrome)	14.90	AR	HSAN3 223900	9q31	***IKBKAP*** *603722*	inhibitor of kappaB kinase complex associated protein	Blumenfeld et al. (1993) Anderson et al. (2001) Slaugenhaupt et al. (2001)	
Hereditary sensory and autonomic neuropathy type IV	14.91	AR	HSAN4 (CIPA) 256800	1biq23.1	***NTRK1*** 191315	neurotrophic receptor tyrosine kinase 1	Indo et al. (1996)	
Hereditary sensory and autonomic neuropathy type V	14.92	AR	HSAN5	1p13.1	***NGFB*** *162030*	nerve growth factor (beta polypeptide)	Einarsdottir et al. (2004)	
Hereditary sensory and autonomic neuropathy type VI	14.93	AR	HSAN6 614653	6p12.1	***DST*** 113810	dystonin	Edvardson et al. (2012)	
Neuropathy, hereditary sensory and autonomic, type VII	14.94	AD	HSAN7 615548	3p22.2	***SCN11A*** 604385	sodium voltage-gated channel alpha subunit 11	Leipold et al. (2013)	
Hereditary sensory and autonomic neuropathy type VIII	14.95	AR	HSAN8 616488	9q34.12	***PRDM12***	PR/SET domain 12 (positive regulatory domain zinc finger protein 12)	Chen et al. (2015)	
Hereditary motor and sensory neuropathy-Lom (with deafness)	14.96	AR	HMNSL 601455	8q24	***NDRG1*** *605262*	Nmyc downstream regulated gene 1	Kalaydjieva et al. (1996, 2000) Hunter et al. (2003)	Allelic to CMT4D (group14)
Hereditary sensory neuropathy with dementia and hearing loss.	14.97	AD	HSN1E 614116	19p13.2	***DNMT1*** *126375*	DNA (cytosine-5)-methyltransferase 1	Klein et al. (2011)	
Peripheral neuropathy and agenesis of the corpus callosum (Charlevoix disease)	14.98	AR	ACCPN 218000	15q13-q14	***SLC12A6 (KCC3)*** *604878*	solute carrier family 12 (potassium chloride cotransporter)	Casaubon et al. (1996) Howard et al. (2002a, 2002b)	
Peripheral neuropathy and deafness, autosomal dominant	14.99	AD		1p34.3	***GJB3*** *603324*	Gap junction protein, beta 3, 31 kDa (=connexin 31)	Lopez-Bigas et al. (2001)	

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Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy)	14.100	AD	HNA 162100	17q25	***SEPT9*** *604061*	septin 9	Pellegrino et al. (1996) Kuhlenbaumer et al. (2005)	
Giant axonal neuropathy	14.101	AR	GAN 256850	16q24.1	***GAN1*** *605379*	gigaxonin	Ben Hamida et al. (1997) Bomont et al. (2000)	
Giant axonal neuropathy 2	14.102	AD	GAN2 610100	1q23.2	***DCAF8*** *615820*	DDB1 and CUL4 associated factor 8	Klein et al. (2014)	
Congenital cataracts, facial dysmorphism and neuropathy	14.103	AR	CCFDN 604168	18p23	***CTDP1*** *604927*	CTD phosphatase subunit 1	Varon et al. (2003)	
Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis	14.104	AR		14q32.2	***VRK1*** *602168*	Vaccinia-related kinase 1	Gonzaga-Jauregui et al. (2013)	
Neuropathy, hereditary sensory, with spastic paraplegia, autosomal recessive	14.105	AR	256840	5p15.2	***CCT5*** *610150*	chaperonin-containing TCP1 subunit 5	Bouhouche et al. (2006a; 2006b)	
GROUP 15. HEREDITARY PARAPLEGIAS See online version of the gene table at http://www.musclegenetable.fr								
GROUP 16. OTHER NEUROMUSCULAR DISORDERS								
Torsion dystonia, early onset	16.1	AD	EOTD 128100	9q34	***TOR1A (DYT1)*** *605204*	Torsin A	Ozelius et al. (1997) Ikeuchi et al. (1999)	
Myoclonus-dystonia syndrome	16.2	AD	DYT11 159900	7q21	***SGCE*** *604149*	Epsilon-sarcoglycan	Klein et al. (2000), Zimprich et al. (2001), Tezenas du Montcel et al. (2006)	
Familial dysautonomia (Riley-Day syndrome)	16.3	AR	HSAN3 223900	9q31	***IKBKAP*** *603722*	Inhibitor of kappab kinase complex associated protein	Blumenfeld et al. (1993), Anderson et al. (2001), Slaugenhaupt et al. (2001)	
Familial amyloid neuropathy	16.4	AD		18q12.1	***TTR*** *176300*	Transthyretin (prealbumin)	Costa et al. (1978) Tawara et al. (1983), Saraiva et al. (1995)	
Congenital fibrosis of the extraocular muscles	16.5	AD	CFEOM1 135700	12q12	***KIF21A*** *608283*	Kinesin family member 21a	Engle et al. (1994), Yamada et al. (2003), Tiab et al. (2004)	
Congenital fibrosis of the extraocular muscles	16.6	AD	CFEOM2 602078	11q13	***PHOX2A (ARIX)*** *602753*	Paired-like aristaless homeobox protein 2a)	Wang et al. (1998) Nakano et al. (2001)	
Congenital fibrosis of the extraocular muscles	16.7	AD	CFEOM3 600638	16q24	***TUBB3*** *602661*	Tubulin, beta 3	Doherty et al. (1999), Tischfield et al. (2010)	
Distal arthrogryposis type 1	16.8	AD	DA1 108120	9p13	***TPM2*** *190990*	Tropomyosin 2, b tropomyosin	Sung et al. (2003a)	Allelic to NEM 4 (group 3), CAP disease (group 3.), DA2B (group 16)
Distal arthrogryposis type 2a, Freeman-Sheldon syndrome	16.9	AD	DA2A 193700	17p13	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al. (2006)	
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.10	AD	DA2B, 601680	11p15	***TNNI2*** *191043*	Troponin i, fast-twich skeletal muscle isoform	Sung et al. (2003a); Kimber et al. (2006)	
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.11	AD	DA2B, 601680	11p15	***TNNT3*** *600692*	Troponin t3, fast skeletal	Sung et al. (2003b)	
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.12	AD	DA2B, 601680	17p13	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al. (2006a)	
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.13	AD	DA2B, 601680	9p13	***TPM2*** *190990*	Tropomyosin 2 (beta)	Tajsharghi et al. (2007c); Ochala et al. (2007)	NEM4 (group 3), CAP disease (group 3), DA1 (group 16)
Arthrogryposis multiplex congenita with nesprin-1 defect	16.14	AR	AMC	6q25	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Attali et al. (2009)	
Arthrogryposis multiplex congenita with cerebral and cerebellar atrophy	16.15	AR	LCCS7 616286	17q21.2	***CNTNAP1*** *602346*	Contactin-Associated Protein 1	Laquérriere et al. (2014)	
Trismus-pseudocamptodactyly	16.16	AD	608837	17p13	***MYH8*** *160741*	Myosin heavy chain, 8, skeletal muscle, perinatal	Veugelers et al. (2004); Toydemir et al. (2006b)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1	16.17	AD	PEOA1 157640	15q25	***POLG*** *174763*	Polymerase, DNA, gamma **(M)**	Van Goethem et al. (2001)	

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Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2;	16.18	AD	PEOA2 609283	4q35	***ANT1*** *103220*	Mitochondrial carrier, adenine nucleotide translocator. ant1 **(M)**]	Kaukonen et al. (2000)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3	16.19	AD	PEOA3 609286	10q24	***PEO1 (C10ORF2)*** *606075*	Twinkle, t7 gene 4-like protein with intramitochondrial nucleoid localization **(M)**	Suomalinen et al. (1997) Spelbrink et al. (2001)	Allelic to IOSCA (group 13)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4	16.20	AD	PEOA4 610131	10q24	***POLG2*** *604983*	Polymerase (DNA directed), gamma 2, accessory subunit **(M)**	Longley et al. (2006)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5	16.21	AD	PEOA5 613077	8q23	***RRM2*** *604712*	Ribonucleotide reductase M2 B **(M)**	Tyynismaa et al., 2009	Allelic to MTDP8B (group 16)
Mitochondrial DNA depletion myopathy	16.22	AR	MTDPS3 609560	16q22	***TK2*** *188250*	Thymidine kinase, mitochondrial **(M)**	Saada et al. (2001)	
Mitochondrial DNA depletion myopathy, encephalomyopathic form	16.23	AR	MTDPS5 612073	13q12-q13	***SUCLA2*** *603921*	Succinate-CoA ligase, adp-forming, beta subunit **(M)**	Elpeleg et al. (2005)	
Mitochondrial DNA depletion myopathy	16.24	AR	MTDP8B 612075	8q23	***RRM2B*** *604712*	Ribonucleotide reductase, M2B **(M)**	Bourdon et al. (2007)	Allelic to PËOA5 (group 16)
Mitochondrial myopathy	16.25	AR		19p13.11	***SLC25A42*** *610823*	solute carrier family 25 member 42 **(M)**	Shamseldin et al. (2015)	
Progressive external ophthalmoplegia with optic atrophy, optic atrophy 1 with deafness	16.26	Ad	125250	3q28-q29	***OPA1*** *605290*	Opa1 protein, dynamin related gtpase **(M)**	Amati-Bonneau et al. (2008) Hudson et al. (2008)	
Tubular aggregate myopathy 1	16.27	AD	TAM1 160565	11p15.4	***STIM1*** *605921*	Stromal interaction molecule 1	Bohm et al. (2013) Hedberg et al. (2014) Nesin et al. (2014),	
Tubular aggregate myopathy 2	16.28	AD	TAM2 615883	12q24.31	***ORAI1*** *610277*	ORAI calcium release-activated calcium modulator 1	Nesin et al. (2014)	
Mitochondrial myopathy and sideroblastic anemia 1	16.29	AR	MLASA1 600462	12q24.33	***PUS1*** *608109*	Pseudouridylate synthase 1 **(M)**	Bykhovskaya et al. (2004) Fernandez-Vizarra (2007)	
Mitochondrial myopathy	16.30	AD		22q11	***CHCHD10*** *(=C22ORF16)* *615903*	Coiled-coil-helix -coiled-coil-helix domain containing 10 **(M)**	Ajroud-Driss et al. (2014)	allelic to FTDALS2 and SMAJ (group 12)
Vacuolar myopathy with accumulation of sarcoplasmic reticulum protein aggregates	16.31	?	VMCQA 616231	1q21	***CASQ1*** *114250*	Calsequestrin 1 (fast-twitch, skeletal muscle) **(M)**	Rossi et al. (2014)	
Myopathy, lactic acidosis, and sideroblastic anemia-2	16.32	AR	MLASA2 613561	12p11.21	***YARS2*** *610957*	Tyrosyl-tRNA synthetase 2, mitochondrial **(M)**	Riley et al. (2010)	
poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis	16.33	AD	POIKTMP 615704	11q12.1	***FAM111B*** *615584*	Family With Sequence Similarity 111, Member B	Mercier et al. (2015)	
Combined Oxidative phosphorylation Deficiency 6;	16.34	XL	COXPD6 300816	Xq26.1	***AIFM1*** *300169*	Apoptosis-Inducing Factor, Mitochondria-Associated, 1 (M)	Morton et al. (2017)	
Mitochondrial myopathy with severe neurological manifestations	16.35	AR		8q24.13	***TMEM65*** *616609*	Transmembrane Protein 65	Nasli et al. (2017)	
MELAS-like syndrome	16.36	AR	220110	2q33.3	***FASTKD2*** *612322*	Fast Kinase Domains 2	Yoo et al. (2017)	
Infantile-onset multisystem disease with progressive muscle weakness	16.37	AR	IMNEPD 616263	17q23.1	***PTRH2*** *608625*	Peptidyl-tRNA Hydrolase 2	Hu et al. (2014)	

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