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The 2022 version of the gene table of neuromuscular disorders (nuclear genome)

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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 neuromuscular 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. *SMA & 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-genetic 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

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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.)*

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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.

Overview of the new data in the 2022 printed version of the gene table (pages 1313 to 1357 of this issue)

There are 39 new items, marked by background shading. Altogether they comprise **25 additional genes,** and **14 additional phenotypic variants** caused by a gene already listed in the 2021 version, one being for an identified locus (item #14.13) (see box).

The new key references of the printed version of the table are listed on pages 1314 in this issue.

As a reminder, we implemented the revised nomenclature of LGMD (group 1) proposed by Straub et al. (2018), keeping the previous nomenclature in parallel in order to allow a smooth transition for users. For CMT (group 14), we decided not to implement the proposed revised nomenclature (Maguy et al. 2018) in the present released printed version of the gene table of neurouscular disorders, in order to allow further time for the neuromuscular community to fully validate these proposed nomenclatures.

Citation of the gene table

- Printed version: Cohen E, Bonne G, Rivier F, Hamroun D. The 2022 version of the gene table of neuromuscular disorders. Neuromuscul Disord. 31 (12), 1313–1357.
- Online version: GeneTable of Neuromuscular Disorders: http://www.musclegenetable.fr

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 groups 12 and 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.

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New in the 2022 printed version of the gene table

25 genes added:

ADPRHL2 (item # 12.105)
APOO (item # 16.81)
CADM3 (item # 14.69)
CHD8 (item # 11.42)
COX16 (item # 16.87)
COX20 (item # 12.103)
GFER (item # 16.84)
GGPS1 (item # 1.17)
JAG2 (item # 1.14)
LIG3 (item # 16.86)
LRIF1 (item # 1.12)
LRP10 (item # 12.76)
MCOLN1 (item # 3.67)
MICU1 (item # 5.37)
MYLPF (item # 16.82)
MYOD1 ((item # 16.83)
NARS1 (item # 14.130)
NSUN3 (item # 16.85)
PLIN4 (item # 4.28)
PSAT1 (item # 12.104)
RFC1 (item # 14.119)
SMPX (item # 4.27)
TMEM126B (item # 16.80)
TNNC2 (item # 3.68)
VWA1 (item # 12.101)

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

ANO5 (item # 1.16)
ANXA11 (item # 12.102)
B4GALNT1 (item # 14.96)
CHRNB1 (item # 11.6)
GBF1 (item #14.13)
GOSR2 (item # 1.15)
MCM3AP (item # 14.95)
MYH7 (item # 3.37)
NOTCH2NLC (item # 5.38)
SPTLC1 (item # 12.77)
STIM1 (item # 5.39)
SYT2 (item # 11.15)
TNNT3 (item # 3.14)
TOR1AIP1 (item # 11.41)

46 new key references

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Gene table of monogenic neuromuscular disorders (nuclear genome only)

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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-p21.1	***DMD*** *300377*	Dystrophin	Monaco et al. (1986) Burghes et al. (1987) Koenig et al. (1987, 1988) Hoffman et al. (1987, 1988)	allelic to CMD3B (group10)
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 6, X-linked	1.3	XR	EDMD6 300696	Xq26.3	***FHL1*** *300163*	Four and a half LIM domain 1	Gueneau et al. (2009)	allelic to XMPMA (group 5), SPM (group 5), RBMX1A/B (group 5)
Emery-Dreifuss muscular dystrophy 2, autosomal dominant	1.4	AD	EDMD2 181350	1q22	***LMNA*** *150330*	Lamin A/C	Bonne et al. (1999) Worman and Bonne (2007)	allelic to EDMD3 (group 1), formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370]
Emery-Dreifuss muscular dystrophy 3, autosomal recessive	1.5	AR	EDMD3 616516	1q22	***LMNA*** *150330*	Lamin A/C	Raffaele di Barletta et al. (2000) Worman and Bonne (2007)	allelic to EDMD2 (group 1), formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370]
Emery-Dreifuss muscular dystrophy 4, autosomal dominant	1.6	AD	EDMD4 612998	6q25.2	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Zhang et al. (2007)	allelic to DCM related to *SYNE1* (group 10A), SCAR8 (group 13), AMCM (group 16)
Emery-Dreifuss muscular dystrophy 5, autosomal dominant	1.7	AD	EDMD5 612999	14q23.2	***SYNE2*** *608442*	Spectrin repeat containing, nuclear envelope 2 (nesprin-2)	Zhang et al. (2007)	
Emery-Dreifuss muscular dystrophy 7, autosomal dominant	1.8	AD	EDMD7 614302	3p25.1	***TMEM43*** *(=LUMA)* *612048*	Transmembrane protein 43 (=LUMA)	Liang et al. (2011)	allelic to ARVD5 (group 10B)
Myopathy with rigid spine and distal joint contractures (Formerly LGMD2Y)	1.9	AR	MRRSDC 617072	1q25.2	***TOR1AIP1*** *(=LAP1B)* *614512*	Torsin A interacting protein 1 (=Lamin Associated Peptide 1B)	Kayman-Kurekci et al. (2014) Fichtman et al. (2019)	Allelic to CMS (group 11)

(*continued on next page*)

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)
Facio-scapulo-humeral muscular dystrophy, type 1	1.10	AD	FSHD1 158900	4q35	***DUX4**** *606009* *(*inappropriate reactivation)*	Double homeobox 4	Wijmenga et al. (1990–1993) 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)	
Facio-scapulo-humeral muscular dystrophy, type 2	1.11	AD	FSHD2 158901	18p11.32	***SMCHD1**** *614982* *(*causing inappropriate reactivation of DUX4* 606,009)*	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)
Facio-scapulo-humeral muscular dystrophy	1.12	AR	FSHD3 619477	1p13.3	***LRIF1*** *615354*	Ligand-Dependent Nuclear Receptor-Interacting Factor 1	Hamanaka et al. (2020)	
Muscular dystrophy with generalized lipodystrophy	1.13	AD	CGL4 613327	17q21.2	***CAVIN1*** *603198*	Caveolae-associated protein 1	Hayashi et al. (2009)	
Muscular dystrophy	1.14	AR		14q32.33	***JAG2*** *602570*	Jagged 2	Coppens et al. (2021)	
Muscular dystrophy	1.15	AR		17q21.32	***GOSR2*** *604027*	Golgi SNAP receptor complex member 2	Stemmerik et al. (2021)	allelic to Congenital muscular dystrophy with hypoglycosylation of dystroglycan and epilepsy (group 2)
Muscular dystrophy with gnathodiaphyseal dysplasia	1.16	AR		11p14.3	***ANO5*** *(TMEM16E)* *608662*	Anoctamin 5	Shaibani et al. (2021)	allelic to LGMDR12 (group 1) and MMD3 (group 4)
Muscular dystrophy with hearing loss and ovarian insufficiency syndrome	1.17	AR	MDHLO 619518	1q42.3	***GGPS1*** *606982*	Geranylgeranyl Diphosphate Synthase 1	Foley et al. (2020)	
Limb girdle muscular dystrophies, dominant								
LGMDD1 (formerly LGDM1E)	1.18	AD	LGMDD1 (LGMD1E) 603511	7q36.3	***DNAJB6*** *611332*	Hsp40 homologue, subfamily B, number 6	Speer et al. (1999), Sarparanta e al (2012) Harms et al. (2012)	allelic to Rimmed vacuole distal myopathy and distal myopathy (group 4)
LGMDD2 (Formerly LGMD1F)	1.19	AD	LGMDD2 (LGMD1F) 608423	7q32.1	***TNPO3*** *610032*	Transportin 3	Palenzuela et al. (2003) Melià et al. (2013) Torella et al. (2013)	allelic to Congenital Myopathy related to TNPO3 (group 3)
LGMDD3 (Formerly LGMD1G)	1.20	AD	LGMDD3 (LGMD1G) 609115	4q21.22	***HNRNPDL*** *607137*	Heterogeneous nuclear ribonucleoprotein D-like	Starling et al. (2005) Vieira et al. (2014)	
LGMDD4	1.21	AD	LGMDD4 (LGMD1I) 618129	15q15.1	***CAPN3*** *114240*	Calpain-3	Vissing et al. (2016) Martinez-Thompson et al. (2018)	allelic to LGMDR1 (group 1)
LGMDD5	1.22	AD	LGMDD5 (BTHLM1) 158810	21q22.3	***COL6A1*** *120220*	Collagen type VI subunit alpha 1	Jöbsis et al. (1996)	allelic to LGMDR12 (group 1), UCMD1 and BTHLM1 (group 2)
LGMDD5	1.23	AD	LGMDD5 (BTHLM1) 158810	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Jöbsis et al. (1996)	allelic to LGMDR22 (group 1); UCMD1, BTHLM1 and Congenital myosclerosis (group 2)
LGMDD5	1.24	AD	LGMDD5 (BTHLM1) 158810	2q37.3	***COL6A3*** *120250*	Collagen type VI subunit alpha 3	Speer et al. (1996) Bertini et al. (1998) Pan et al. (1998)	allelic to LGMDR22 (group 1); UCMD1 and BTHLM1 (group 2)

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Myofibrillar myopathy 3 (Formerly LGMD1A)	1.25	AD	MFM3 609200 (LGMD1A 159000)	5q31	***MYOT*** *604103*	Myotilin	Speer et al. (1992) Hauser et al. (2000)	allelic to Distal myotilinopathy (group 4), MFM3 (group 5), spheroid body myopathy (group 5)
Emery-Dreifuss muscular dystrophy 2 (Formerly LGMD1B)	1.26	AD	EDMD2 181350 (LGMD1B 159001)	1q22	***LMNA*** *150330*	Lamin A/C	van der Kooi et al. (1997) Muchir et al. (2000) Worman and Bonne (2007)	allelic to EDMD2, EDMD3 and formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
Rippling muscle disease 2 (Formerly LGMD1C)	1.27	AD	RMD2 606072 (LGMD1C 607801)	3p25.3	***CAV3*** *601253*	Caveolin-3	Minetti et al. (1998) McNally et al. (1998)	allelic to MPDT (group 4), Creatin phosphokinase elevated serum (group 5), RMD2 (group 6), CMH1 (group 10A), LQT9 (group 10B)
Myofibrillar myopathy 1 (Formerly LGMD1 related to *DES*)	1.28	AD	MFM1 601419	2q35	***DES*** *125660*	Desmin	Messina et al. (1997) Greenberg et al. (2012) Hedberg et al. (2012)	allelic to formerly LGMD2R (group 1), MFM1 with or without ARCV7 (group 5), CMD1I (group 10A), ARVC7 (group 10B)
LGMD1H	1.29	AD	LGMD1H 613530	3p25.1-p23	*?*	?	Bisceglia et al. (2010)	
Limb girdle muscular dystrophies, recessive								
LGMDR1 (Formerly LGMD2A)	1.30	AR	LGMDR1 (LGMD2A) 253600	15q15.1	***CAPN3*** *114240*	Calpain-3	Beckmann et al. (1991) Young et al. (1992) Richard et al. (1995, 1997)	allelic to LGMDD4 (group 1)
LGMDR2 (Formerly LGMD2B)	1.31	AR	LGMDR2 (LGMD2B) 253601	2p13.2	***DYSF*** *603009*	Dysferlin	Bashir et al. (1994) Bashir et al. (1998) Liu et al. (1998)	allelic to MMD1 (group 4)
LGMDR3 (Formerly LGMD2D)	1.32	AR	LGMDR3 (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)	
LGMDR4 (Formerly LGMD2E)	1.33	AR	LGMDR4 (LGMD2E) 604286	4q12	***SGCB*** *600900*	Beta-sarcoglycan	Lim et al. (1995) Bönnemann et al. (1995) Bönnemann et al. (1996)	
LGMDR5 (Formerly LGMD2C)	1.34	AR	LGMDR5 (LGMD2C) 253700	13q12.12	***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)	
LGMDR6 (Formerly LGMD2F)	1.35	AR	LGMDR6 (LGMD2F) 601287	5q33.3-q33.3	***SGCD*** *601411*	Delta-sarcoglycan	Passos-Bueno et al. (1996) Nigro et al. (1996)	allelic to CMD1L (group 10A)
LGMDR7 (Formerly LGMD2G	1.36	AR	LGMDR7 (LGMD2G) 601954	17q12	***TCAP*** *604488*	Titin-cap (telethonin)	Moreira et al. (1997) Moreira et al. (2000)	allelic to CMD related to *TCAP* (group 2), CMH25 (group 10A), CMD1N (group 10A)
LGMDR8 (Formerly LGMD2H	1.37	AR	LGMDR8 (LGMD2H) 254110	9q33.1	***TRIM32*** *602290*	Tripartite motif-containing 32	Weiler et al. (1998) Frosk et al. (2002)	allelic to sarcotubular myopathy (group 3)
LGMDR9 (Formerly LGMD2I	1.38	AR	LGMDR9 (MDDGC5) 607155	19q13.32	***FKRP*** *606596*	Fukutin related protein	Driss et al. (2000) Brockington et al. (2001a)	allelic to MDDGB5 (group 2), MDDGA5/WWS (group 2), MDDGA5/MEB (group 2)
LGMDR10 (Formerly LGMD2J)	1.39	AR	LGMDR10 (LGMD2J) 608807	2q31.2	***TTN*** *188840*	Titin	Hackman et al. (2003)	allelic to CNM related to *TTN* (group 3), MmD related to *TTN* (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10), CMD1G (group 10), LCCS related to *TTN* (group 12)

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LGMDR11 (Formerly LGMD2K)	1.40	AR	LGMDR11 (MDDGC1) 609308	9q34.13	***POMT1*** *607423*	Protein 0-mannosyltransferase 1	Balci et al. (2005) D'Amico et al. (2006)	allelic to WWS/MDDGA1 (group 2) and MDDGB1 (group 2)
LGMDR12 (Formerly LGMD2L)	1.41	AR	LGMDR12 (LGMD2L) 611307	11p14.3	***ANO5*** *(TMEM16E) 608662*	Anoctamin 5	Jarry et al. (2007) Bolduc et al. (2008, 2010) Hicks et al. (2011)	allelic to Muscular dystrophy with gnathodiaphyseal dysplasia (group 1) and MMD3 (group 4)
LGMDR13 (Formerly LGMD2M)	1.42	AR	LGMDR13 (MDDGC4) 611588	9q31.2	***FKTN*** *607440*	Fukutin	Murakami et al. (2006) Godfrey et al. (2006)	allelic to MDDGA4/FCMD (group 2), MDDGB4/WWS (group 2), CMD1X (group 10A)
LGMDR14 (Formerly LGMD2N)	1.43	AR	LGMDR14 (MDDGC2) 613158	14q24.3	***POMT2*** *607439*	Protein O-mannosyl transferase 2	Biancheri et al. (2007)	allelic to MDDGA2/MDDGB2/WWS/ MEB (group 2)
LGMDR15 (Formerly LGMD2O)	1.44	AR	LGMDR15 (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 MDDGA3/MDDGB3/WWS (group 2) and MDDGA3/MEB (group 2)
LGMDR16 (Formerly LGMD2P)	1.45	AR	LGMDR16 (MDDGC9) 613818	3p21.31	***DAG1*** *128239*	Dystrophin-associated glycoprotein 1 (alpha-dystroglycan)	Hara et al. (2011)	allelic to MDDGA9 (group 2)
LGMDR17 (Formerly LGMD2Q)	1.46	AR	LGMDR17 (LGMD2Q) 613723	8q24.3	***PLEC*** *601282*	Plectin	Gundesli et al. (2010)	allellic to LGMD with ophthalmoplegia (group 1), EBSMD (group 5), and Myasthenic syndrome related to *PLEC* (group 11)
LGMDR18 (Formerly LGMD2S)	1.47	AR	LGMDR18 (LGMD2S) 615356	4q35.1	***TRAPPC11*** *614138*	Trafficking protein particle complex 11	Bögershausen et al. (2013)	allelic to CMD related to *TRAPPC11* (group 2)
LGMDR19 (Formerly LGMD2T)	1.48	AR	LGMDR19 (MDDGC14) 615352	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Carss et al. (2013) Cabrera-Serrano et al. (2015)	allelic to MEB/MDDGA14, MDDGB14 (group 2) and congenital myasthenic syndrome (group 11)
LGMDR20 (Formerly LGMD2U)	1.49	AR	LGMDR20 (MDDGC7) 616052	7p21.2-p21.1	***CRPPA*** *614631*	Isoprenoid synthase domain containing protein	Tasca et al. (2013)	allelic to WWS/ MDDGA7 (group 2)
LGMDR21 (Formerly LGMD2Z)	1.50	AR	LGMDR21 (LGMD2Z) 617232	3q13.33	***POGLUT1*** *615618*	Protein O-Glucosyltransferase 1	Servian-Morilla et al. (2016)	
LGMDR22 (Bethlem myopathy 1)	1.51	AR	LGMDR22 (UCMD1) 254090	21q22.3	***COL6A1*** *120220*	Collagen type VI subunit alpha 1	Pan et al. (2003) Giusti et al. (2005)	allelic to LGMDD5 (group 1), UCMD1 and BTHLM1 (group 2)
LGMDR22 (Bethlem myopathy 1)	1.52	AR	LGMDR22 (UCMD1) 254090	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Jokela et al. (2019)	allelic to LGMDD5 (group 1), UCMD1, BTHLM1 and congenital myosclerosis (group 2)
LGMDR22 (Bethlem myopathy 1)	1.53	AR	LGMDR22 (UCMD1) 254090	2q37.3	***COL6A3*** *120250*	Collagen type VI subunit alpha 3	Demir et al. (2002)	allelic to LGMDD5 (group 1); UCMD1 and BTHLM1 (group 2)
LGMDR23	1.54	AR	LGMDR23 618138	6q22.33	***LAMA2*** *156225*	Laminin 2, Heavy chain (laminin alpha2 chain of merosin)	Gavassini et al. (2011)	allelic to MDC1A (group 2)
LGMDR24	1.55	AR	LGMDR24 (MDDGC8) 618135	3p22.1	***POMGNT2*** *614828*	Protein O-mannose beta-1,4-N-acetylglucosaminyl transferase 2	Endo et al. (2015)	allelic to MDDGA8 (group 2)
LGMDR25 (Formerly LGMD2X)	1.56	AR	LGMDR25 (LGMD2X) 616812	6q21	***BVES*** *(= POPDC1) 604577*	Blood vessel epicardial substance	Schindler et al. (2016)	
LGMDR26	1.57	AR	LGMDR26 618848	6q21	***POPDC3*** *605824*	Popeye domain-containing protein 3	Vissing et al. (2019)	
Myofibrillar myopathy 1 (Formerly LGMD2R)	1.58	AR	MFM1 601419 (LGMD2R 615325)	2q35	***DES*** *125660*	Desmin	Cetin et al. (2013)	allelic to formerly LGMD1 related to *DES* (group 1), MFM1 with or without ARCV7 (group 5), CMD1I (group 10A) and ARVC7 (group 10B)
Pompe disease (Formerly LGMD2V)	1.59	AR	GSD2 232300	17q25.3	***GAA*** *606800*	Glucosidase alpha, acid	Preisler et al. (2013)	allelic to GSD2 (groups 9 and 10A)

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Muscular dystrophy with cardiomyopathy and triangular tongue (Formerly LGMD2W)	1.60	AR	MDRCMTT (LGMD2W) 616827	2q14.3	***LIMS2*** *(= PINCH2)* *607908*	LIM and senescent cell antigen-like domains 2	Chardon et al. (2015)	
Muscle dystrophy with glycosylation defect, type Io	1.61	AR	MDDGC15 (CDG1O) 612937	1q22	***DPM3*** *605951*	Dolichyl-phosphate mannosyltransferase polypeptide 3	Lefeber et al. (2009)	
Scapuloperoneal muscular dystrophy and dropped head syndrome	1.62	AR	600416	9p13.3	***VCP*** *601023*	Valosin-containing protein	Liewluck et al. (2014)	allelic to IBMPFD1 (groups 4 and 5), ALS14 (group 12) and CMT2Y (group 14)
LGMD with ophthalmoplegia	1.63	AR		8q24.3	***PLEC*** *601282*	Plectin	Fattahi et al. (2015)	allelic to LGMDR17 (group 1), EBSMD (group 5), myasthenic syndrome related to *PLEC* (group 11)
LGMD related to PYROXD1	1.64	AR		12p12.1	***PYROXD1*** *617220*	Pyridine nucleotide-disulphide oxidoreductase domain 1	Sainio et al. (2018)	allelic to Congenital Myopathy related to *PYROXD1* (group 3). MMF8 (group 5)
LGMD related to KBTBD13	1.65	AD		15q22.31	***KBTBD13*** *613727*	Kelch repeat and BTB/POZ domain containing protein 13	Garibaldi et al. (2018)	allelic to NEM6 (group 3)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C12	1.66	AR	MDDGC12 616094	8p11.21	***POMK*** *615247*	Protein-O-mannose kinase	Di Costanzo et al. (2014)	Allelic to MDDGA12 (group 2)
GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES								
Congenital muscular dystrophy with merosin deficiency	2.1	AR	MDC1A 607855	6q22.33	***LAMA2*** *156225*	Laminin 2, Heavy chain (laminin alpha2 chain of merosin)	Tomé et al. (1994) Hillaire et al. (1994) Helbling-Leclerc et al. (1995) Allamand et al. (1997)	allelic to LGMDR23 (group 1)
Bethlem myopathy 1	2.2	AD	BTHLM1 158810	21q22.3	***COL6A1*** *120220*	Collagen type VI subunit alpha 1	Jöbsis et al. (1996)	allelic to LGMDD5 and LGMDR22 (group 1) and UCMD1 (group 2)
Bethlem myopathy 1	2.3	AD	BTHLM1 158810	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Jöbsis et al. (1996)	allelic to LGMDD5 and LGMDR22 (group 1) and UCMD1 and Congenital myosclerosis (group 2)
Bethlem myopathy 1	2.4	AD	BTHLM1 158810	2q37.3	***COL6A3*** *120250*	Collagen type VI subunit alpha 3	Speer et al. (1996) Bertini et al. (1998) Pan et al. (1998)	allelic to LGMDD5 and LGMDR22 (group 1); UCMD1 (group 2)
Bethlem myopathy 1	2.5	AR	BTHLM1 158810	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Gualandi et al. (2009)	allelic to LGMDD5 and LGMDR22 (group 1) and UCMD1 and Congenital myosclerosis (group 2)
Ullrich congenital muscular dystrophy 1	2.6	AR	UCMD1 254090	21q22.3	***COL6A1*** *120220*	Collagen, type VI, subunit alpha 1	Pan et al. (2003) Giusti et al. (2005)	allelic to LGMDD5 and LGMDR22 (group 1) and BTHLM1 (group 2)
Ullrich congenital muscular dystrophy 1	2.7	AR	UCMD1 254090	21q22.3	***COL6A2*** *120240*	Collagen, type VI, subunit alpha 2	Vanegas et al. (2001) Higuchi et al. (2001)	allelic to LGMDD5 and LGMDR22 (group 1); BTHLM1 and congenital myosclerosis (group 2)
Ullrich congenital muscular dystrophy 1	2.8	AR	UCMD1 254090	2q37.3	***COL6A3*** *120250*	Collagen type VI subunit alpha 3	Demir et al. (2002)	allelic to LGMDD5 and LGMDR22 (group 1) and BTHLM1 (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)	Allelic to BTHLM2 (group 2), CMD related to COL12A1 (group 2)
Bethlem myopathy 2	2.10	AD	BTHLM2 *616471*	6q13-q14	***COL12A1*** *120320*	Collagen type XII alpha 1 chain	Zou et al. (2014)	Allelic to UCMD2 (group 2), CMD related to COL12A1 (group 2)
COL12A1-related congenital muscular dystrophy	2.11	AD		6q13-q14	***COL12A1*** *120320*	Collagen type XII alpha 1 chain	Punetha et al. (2016)	Allelic to UCMD2 (group 2), BTHLM2 (group 2)
Myosclerosis, congenital	2.12	AR	255600	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Merlini et al. (2008)	allelic to LGMDD5 and LGMDR22 (group 1); UCMD1 and BTHLM1 (group 2)

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Rigid spine syndrome 1	2.13	AR	RSMD1 602771	1p36.11	***SELENON*** *(formerly SEPN1)* *606210*	Selenoprotein N	Moghadaszadeh et al. (1998, 2001) Ferreiro et al. (2002, 2004)	allelic to CFTD (group 3), multiminicore disease (group 3), and desmin-related myopathy with Mallory bodies (group 5)
Congenital muscular dystrophy due to ITGA7 deficiency	2.14	AR	613204	12q13.2	***ITGA7*** *600536*	Integrin alpha7	Hayashi et al. (1998)	
Congenital muscular dystrophy related to *DNM2*	2.15	AD		19p13.2	***DNM2*** *602378*	Dynamin 2	Susman et al. (2010)	allelic to CNM1 (group 3), Distal myopathy related to *DNM2* (group 4), LCCS5 (group 12) and CMTDIB (group 14)
Congenital musuclar dystrophy related to *TCAP*	2.16	AR		17q12	***TCAP*** *604488*	Titin-cap (telethonin)	Ferreiro et al. (2011)	allelic to LGMDR7 (group 1), CMH25 (group 10A), CMD1N (group 10A)
Congenital muscular dystrophy related to *LMNA*	2.17	AD	MDCL 613205	1q22	***LMNA*** *150330*	Lamin A/C	Quijano-Roy et al. (2008)	allelic to EDMD2, EDMD3 and formerly LGMD1B (group 1), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151,660, HGPS #176,670, restrictive dermopathy #275,210, MADA #248370]
CMD-Dystroglycanopathies								
Fukuyama congenital muscular dystrophy (FCMD)	2.18	AR	MDDGA4 253800	9q31.2	***FKTN*** *607440*	Fukutin	Toda et al. (1993) Kobayashi et al. (1998)	allelic to LGMDR13 (group 1), WWS/MDDGB4 (group 2), CMD1X (group 10A)
Walker-Warburg syndrome (WWS)	2.19	AR	MDDGB4 613152	9q31.2	***FKTN*** *607440*	Fukutin	Beltran-Valero de Bernabe (2003) Mercuri et al. (2009)	allelic to LGMDR13 (group 1) and FCMD/MDDGA4 (group 2) and CMD1X (group 10A)
Walker-Warburg syndrome (WWS)	2.20	AR	MDDGA1 236670	9q34.13	***POMT1*** *607423*	Protein-O-mannosyl transferase 1	Beltran-Valero De Bernabe et al. (2002) Mercuri et al. (2009)	allelic to LGMDR11 (group 1), and MDDGB1 (group 2)
Walker-Warburg syndrome (WWS)	2.21	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 LGMDR14 (group 1) and MDDGA2/MDDGB2/MEB (group 2)
Walker-Warburg syndrome (WWS)	2.22	AR	MDDGA5 613153	19q13.32	***FKRP*** *606596*	Fukutin related protein	Beltran-Valero De Bernabe et al. (2004)	allelic to LGMDR9 (group 1), MDDGB5 (group 2), MDDGA5/MEB (group 2)
Walker-Warburg syndrome (WWS)	2.23	AR	MDDGA3 253280 MDDGB3 613151	1p34.1	***POMGNT1*** *606822*	Protein O-mannose beta1,2-N-acetylglucosaminyl transferase	Taniguchi et al. (2003) Mercuri et al. (2009)	allelic to LGMDR15 (group 1) and MDDGA3/MEB (group 2)
Walker-Warburg syndrome (WWS)	2.24	AR	MDDGA7 614643	7p21.2	***CRPPA*** *614631*	Isoprenoid synthase domain containing	Roscioli et al. (2012) Willer et al. (2012)	Alletic to LGMDR20 (group 1)
Walker-Warburg syndrome (WWS)	2.25	AR	MDDGA8 614830	3p22.1	***POMGNT2*** *614828*	Protein O-mannose beta-1,4-N-acetylglucosaminyl transferase 2	Manzini et al. (2012)	allelic to LGMDR24 (group 1)
Walker-Warburg syndrome (WWS)	2.26	AR	MDDGA13 615287	11q13.2	***B4GAT1*** *605517*	Beta-1,4-glucuronyl transferase 1 (Beta-1,3-N-acetylglucosaminyl-transferase 1)	Buysse et al. (2013) Shaheen et al. (2013)	
Muscle-eye-brain disease (MEB)	2.27	AR	MDDGA3 253280	1p34.1	***POMGNT1*** *606822*	Protein O-mannose beta1,2-N-acetylglucosaminyl transferase	Yoshida et al. (2001) Taniguchi et al. (2003)	allelic to LGMDR15 (group 1) and MDDGA3/WWS (group 2)
Muscle-eye-brain disease (MEB)	2.28	AR	MDDGA5 613153	19q13.32	***FKRP*** *606596*	Fukutin related protein	Beltran-Valero De Bernabe et al. (2004)	allelic to LGMDR9 (group 1), MDDGB5 (group 2), MDDGA5/WWS (group 2)
Muscle-eye-brain disease (MEB)	2.29	AR	MDDGA2 613150 MDDGB2 613156	14q24.3	***POMT2*** *607439*	Protein O-mannosyl transferase 2	Mercuri et al. (2006)	allelic to LGMDR14 (group 1) and MDDGA2/MDDGB2/WWS (group 2)
Muscle-eye-brain disease (MEB)	2.30	AR	MDDGA14 615350	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	de Carss et al. (2013)	allelic to LGMDR19 (group 1), MDDGB14 (group 2), congenital myasthenic syndrome (group 11)

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Muscle-eye-brain disease (MEB)	2.31	AD	MEB	11q13.2q14.1	***11q13.2q14.1 duplication***		Villar-Quiles et al. (2020)	
Muscular dystrophy-dystrooglycanopathy (congenital with mental retardation), type B1	2.32	AR	MDDGB1 607423	9q34.13	***POMT1*** *607423*	Protein-O-mannosyl transferase 1	van Reeuwijk et al. (2006) Mercuri et al. (2009)	allelic to LGMDR11 (group 1) and MDDGA1 (group 2)
Muscular dystrophy-dystroglycanopathy, congenital with or without mental retardation (formerly MDC1C)	2.33	AR	MDDGB5 606612	19q13.32	***FKRP*** *606596*	Fukutin related protein	Brockington et al. (2001b) Topaloglu et al. (2003) Mercuri et al. (2009)	allelic to LGMDR9 (group 1), MDDGA5/WWS (group 2), MDDGA5/MEB (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.34	AR	MDDGA6 613154 MDDGB6 608840	22q12.3	***LARGE1*** *603590*	Acetylglucosaminyl-transferase-like protein (like-glycosyl transferase)	Longman et al. (2003) Mercuri et al. (2009)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.35	AR	CDG1E 608799	20q13.13	***DPM1*** *603503*	Dolichyl-phosphate mannosyltransferase 1, catalytic subunit	Yang et al. (2013)	
Congenital disorder of glycosylation type 1	2.36	AR	CDG1F 609180	17p13.1	***MPDU1*** *604041*	Mannose-P-dolichol utilization defect 1	van Tol et al. (2019)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and severe epilepsy	2.37	AR	CDG1U 615042	9q34.11	***DPM2*** *603564*	Dolichyl-phosphate mannosyltransferase polypeptide 2, regulatory subunit	Barone et al. (2012)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.38	AR	MDDGA9 616538	3p21.31	***DAG1*** *128239*	Dystrophin-Associated Glycoprotein 1	Geis et al. (2013)	allelic to LGMDR16 (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A10	2.39	AR	MDGGA10 615041	12q14.2	***RXYLT1*** *605862*	Ribitol xylosyltransferase 1 (transmembrane protein 5)	Vuillaumier-Barrot et al. (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan WWWS/MEB like	2.40	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	2.41	AR	MDDGA12 615249	8p11.21	***POMK*** *615247*	Protein-O-mannose kinase	Jae et al. (2013)	Allelic to MDDGC12 (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and mental retardation	2.42	AR	MDDGB14 615351	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Carss et al. (2013)	allelic to LGMDR19 (group 1), MEB/MDDGA14 (group 2) and congenital myasthenic syndrome (group 11)
Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by *TRAPPC11*mutations	2.43	AR		4q35.1	***TRAPPC11*** *614138*	Trafficking protein particle complex 11	Liang et al. (2015) Larson et al. (2018)	allelic to LGMDR18 (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and epilepsy	2.44	AR		17q21.32	***GOSR2*** *604027*	Golgi SNAP receptor complex member 2	Larson et al. (2018)	
Other congenital muscular dystrophies								
Congenital muscle dystrophy with joint hyperlaxity	2.45	AR		3p23–21	***?***		Tetreault et al. (2006)	
Congenital muscle dystrophy with mitochondrial structural abnormalities (megaconial type)	2.46	AR	MDCMC 602541	22q13.33	***CHKB*** *612395*	Choline kinase beta	Mitsuhashi et al. (2011)	
Congenital muscular dystrophy	2.47	AR	MDC1B 604801	1q42	***?***		Brockington et al. (2000)	

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Congenital muscular dystrophy with rigid spine related to *ACTA1*	2.48	AR	Possibly identical to MDC1B 604801	1q42.13	***ACTA1*** *102610*	Alpha actin, skeletal muscle	O'Grady et al. (2014)	allelic to NEM3, CFTD, Cap myopathy (group 3)
GOLGA2-related congenital muscle dystrophy with brain involvement	2.49	AR		9q34.11	***GOLGA2*** *602580*	Golgin A2	Shamseldin et al. (2016)	
Muscular dystrophy, congenital Davignon-Chauveau type	2.50	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	2.51	AR	MDCCAID 617404	17p13.3	***INPP5K*** *607875*	Inositol Polyphosphate-5-Phosphatase K	Osborn et al. (2017) Wiessner et al. (2017)	
Congenital muscular dystrophy related to *MSTO1*	2.52	AR		1q22	***MSTO1*** *617619*	Misato 1, mitochondrial distribution and morphology regulator (**M**)	Nasca et al. (2017) Ardicli et al. (2019) Donkervoort et al. (2019)	allelic to MMYAT (group 13 and 16)
Congenital muscular dystrophy related to *RYR1*	2.53	AR		19q13.2	***RYR1*** *180901*	Ryanodine receptor	Helbling et al. (2019)	allelic to CFTD, CNM related to RYR1, CCD, CNMDU1, minicore myopathy with external ophthalmoplegia, DuCD (group 3), Late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to *RYR1* (group 16)
GROUP 3. CONGENITAL MYOPATHIES								
Nemaline myopathies								
Nemalin myopathy 1	3.1	AD	NEM1 609284	1q21.3	***TPM3*** *191030*	Tropomyosin 3	Laing et al. (1992) Laing et al. (1995b) Tan et al. (1999) Wattanasirichaigoon et al. (2002)	allelic to CFTD, CAPM1 (group 3)
Nemalin myopathy 2	3.2	AR	NEM2 256030	2q23.3	***NEB*** *161650*	Nebulin	Wallgren-Pettersson et al. (1995, 2002) Pelin et al. (1999) Lehtokari et al. (2006)	allelic to Distal myopathy with nebulin defect (group 4)
Nemalin myopathy 3	3.3	AD, AR	NEM3 161800	1q42.13	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Nowak et al. (1999) Sparrow et al. (2003)	allelic to CMD with rigid spine (group 2), CFTD and cap myopathy (group 3)
Nemalin myopathy 4	3.4	AD	NEM4 609285	9p13.3	***TPM2*** *190990*	Tropomyosin 2 (beta)	Donner et al. (2002) Monnier et al. (2009)	allelic to CAPM2 (group 3), DA2B and DA1A (group 16)
Nemalin myopathy 5	3.5	AR	NEM5 605355	19q13.42	***TNNT1*** *191041*	Troponin T type 1 (skeletal, slow)	Johnston et al. (2000)	
Nemalin myopathy 6	3.6	AD	NEM6 609273	15q22.31	***KBTBD13*** *613727*	Kelch repeat and BTB/POZ domain containing protein 13	Gommans et al. (2003) Samburghin et al. (2010)	allelic to LGMD related to *KBTBD13* (group 1)
Nemalin myopathy 7	3.7	AR	NEM7 610687	14q13.1	***CFL2*** *601443*	Cofilin 2 (muscle)	Agrawal et al. (2007)	
Nemalin myopathy 8	3.8	AR	NEM8 615348	3p22.1	***KLHL40*** *615340*	Kelch-like family member 40	Ravenscroft et al. (2013)	
Nemalin myopathy 9	3.9	AR	NEM9 615731	2q31.1	***KLHL41*** *607701*	Kelch-like family member 41	Gupta et al. (2013)	
Nemalin myopathy 10	3.10	AR	NEM 10 616165	3p14.1	***LMOD3*** *616112*	Leiomodin 3	Yuen et al. (2014)	
Nemalin myopathy 11	3.11	AR	NEM11 617336	10q21.3	***MYPN*** *608517*	Myopalladin	Miyatake et al. (2016) Lornage et al. (2017)	allelic to CMD1KK, CMH22 and RCM4 (group 10A)
Klippel-Feil syndrome with Nemalin myopathy and facial dysmorphism	3.12	AR	KFS4 616549	22q12.1	***MYO18B*** *607295*	Myosin XVIIIB	Malfatti et al. (2015)	
Myopathy with nemaline bodies	3.13	AR		15q13-q14	***RYR3*** *180903*	Ryanodine receptor 3	Nilipour et al. (2018)	

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Nemalin myopathy with distal arthrogryposis	3.14	AR		11p15.5	***TNNT3*** *600692*	Troponin T3, fast skeletal	Sandaradura et al. (2018)	Allelic to DA2B2 (group 16)
Other congenital myopathies								
Myopathy, congenital, with fiber-type disproportion 1	3.15	AD, AR	CFTD 255310	1q42.13	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Clarke et al. (2003) Laing et al. (2004)	allelic to CMD with rigid spine (group 2), NEM3 and cap myopathy (group 3)
Myopathy, congenital, with fiber-type disproportion	3.16	AD, AR	CFTD 255310	1p36.11	***SELENON*** *(formerly SEPN1) 606210*	Selenoprotein N	Clarke et al. (2006)	allelic to RSMD1 (group 2), CFTD (group 3), Multminicore disease (group 3), *DES*-related myopathy with Mallory bodies (group 5)
Myopathy, congenital, with fiber-type disproportion	3.17	AD, AR	CFTD 255310	1q21.3	***TPM3*** *191030*	Tropomysosin 3	Clarke et al. (2008)	allelic to NEM1, CAPM1 (group 3)
Myopathy, congenital, with fiber-type disproportion	3.18	AR	CFTD 255310	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Clarke et al. (2010)	allelic to CMD related to *RYR1* (group 2), CNM related to *RYR1*, CCD, CNMDU1, minicore myopathy with external ophthalmoplegia (group 3), Late onset axial myopathy (group 5), MHS1 (group 8) Fetal akinesia related to *RYR1* (group 16)
Myopathy congenital, with fiber-type disproportion	3.19	AD	CFTD 255310	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Ortolano et al. (2011)	allelic to MSMA, MSMB, eccentric core disease (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10A)
Myopathy congenital, with fiber-type disproportion	3.20	AR	CFTD 255310	12q24.11	***MYL2*** *160781*	Myosin regulatory ligh chain 2	Waterman et al. (2013)	Allelic to CMH10 (group 10A)
Myopathy congenital, with fiber-type disproportion	3.21	AR	CNM6 617760	2q31.1	***ZAK*** *609479*	Leucine zipper-and sterile alpha motif-containing kinase	Vasli et al. (2017)	
Myotubular myopathy, X-linked	3.22	XR	CNMX 310400	Xq28	***MTM1*** *300415*	Myotubularin 1	Thomas et al. (1987) Laporte (1996, 1997, 2000)	
Centronuclear myopathy 1	3.23	AD	CNM1 160150	19p13.2	***DNM2*** *602378*	Dynamin 2	Bitoun et al. (2005)	allelic to CMD related to *DNM2* (group 2), CNM1 (group 4), LCCS5 (group 12) and CMTDIB (group 14)
Centronuclear myopathy 2	3.24	AR, AD	CNM2 255200	2q14.3	***BIN1*** *601248*	Bridging integrator 1 (Amphiphysin)	Nicot et al. (2007) Böhm et al. (2014)	
Centronuclear myopathy related to *RYR1*	3.25	AR		19q13.2	***RYR1*** *180901*	Ryanodine receptor	Wilmshurst et al. (2010)	allelic to CMD related to RYR1 (group 2), CFTD, CCD, minicore myopathy with external ophthalmoplegia, CNMDU1, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to *RYR1* (group 16)
Centronuclear myopathy (CNM) related to *TTN*	3.26	AR		2q31.2	***TTN*** *188840*	Titin	Ceyhan-Birsoy et al. (2013)	allelic to LGMDR10 (group 1), MmD related to *TTN* (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A), LCCS related to *TTN* (group 12)
Centronuclear myopathy 5	3.27	AR	CNM5 615959	2q35	***SPEG*** *615950*	SPEG complex locus	Agrawal et al. (2014)	
Centronuclear myopathy 4	3.28	AD	CNM4 614807	16p13.3	***CCDC78*** *614666*	Coiled-coil domain-containing protein 78	Majczenko et al. (2012)	
Central core disease, dominant	3.29	AD	CCD 117000	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Kausch et al. (1991) Zhang et al. (1993) Quane et al. (1993) Robinson et al. (2002)	allelic to CMD related to *RYR1* (group 2), CFTD, CNM related to *RYR1*, minicore myopathy with external ophthalmoplegia, CNMDU1, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to *RYR1* (group 16)

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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)
Central core disease, recessive (transient multiminicore myopathy)	3.30	AR	CCD 117000	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Ferreiro et al. (2002a) Jungbluth et al. (2002)	allelic to CMD related to *RYR1* (group 2), CFTD, CNM related to *RYR1*, minicore myopathy with external ophthalmoplegia, CNMDU1 (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to *RYR1* (group 16)
Minicore myopathy with external ophthalmoplegia	3.31	AR	255320	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Monnier et al. (2003) Jungbluth et al. (2005)	allelic to CMD related to *RYR1* (group 2), CFTD, CNM related to *RYR1*, CCD, CNMDU1, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to *RYR1* (group 16)
Multiminicore disease (MmD), classical form	3.32	AR	255320	1p36.11	***SELENON*** *(formerly SEPN1) 606210*	Selenoprotein N	Ferreiro et al. (2002) Ferreiro et al. (2004)	allelic to RSMD1 (group 2), CFTD (group 3), *DES*-related myopathy with Mallory bodies (group 5)
Multiminicore disease (MmD) related to *TTN*	3.33	AR		2q31.2	***TTN*** *188840*	Titin	Ge et al. (2019)	allelic to LGMDR10 (group 1), CNM related to *TTN* (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A), LCCS related to *TTN* (group 12)
Early onset myopathy, areflexia, respiratory distress and dysphagia	3.34	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	Multiple EGF-like-domains 10	Logan et al. (2011)	Allelic to recessive congenital myopathy with minicores (group 3)
Recessive congenital myopathy with minicores	3.35	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	Multiple EGF-like-domains 10	Boyden et al. (2012)	Allelic to EMARDD (group 3)
Myopathy, myosin storage, autosomal dominant	3.36	AD	MSMA 608358	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Tajsharghi et al. (2003) Bohlega et al. (2004) Laing et al. (2005)	allelic to CFTD, MSMB, SPMM, eccentric core disease (group 3); MPD1 (group 4); CMH1, CMD1S and LVNC5 (group 10A)
Myopathy, myosin storage, autosomal dominant (eccentric core disease)	3.37	AD		14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Romero et al. (2014)	allelic to CFTD, MSMA, MSMB, SPMM, (group 3); MPD1 (group 4); CMH1, CMD1S and LVNC5 (group 10A)
Myopathy, myosin storage, autosomal domin	3.38	AR	MSMB 255160	14q11.2	***MYH7*** *160780*	Myosin, heavy chain 7, cardiac muscle, beta	Onengut et al. (2004) Tajsharghi et al. (2007) Yuceyar et al. (2015)	allelic to CFTD, MSMA, SPMM, eccentric core disease (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10A)
Myopathy, proximal, and ophthalmoplegia (inclusion body myopathy 3)	3.39	AD, AR	MYPOP 605637	17p13.1	***MYH2*** *160740*	Myosin, heavy chain 2, skeletal muscle, adult	Martinsson et al. (1999, 2000) Tajsharghi et al. (2010)	
Isolated inclusion body myopathy	3.40	AD	IBMPFD3 615424	12q13.13	***HNRNPA1*** *164017*	Heterogeneous nuclear ribonucleoprotein A1	Izumi et al. (2015)	allelic to ALS20 (group 12)
Cap myopathy CAPM1	3.41	AD, AR	NEM1 609284	1q21.3	***TPM3*** *191030*	Tropomysosin 3	De Paula et al. (2009) Ohlsson et al. (2009)	allelic to NEM1, CFTD (group 3)
Cap myopathy CAPM2	3.42	AD	NEM4 609285	9p13.3	***TPM2*** *190990*	Tropomyosin 2, beta	Tajsharghi et al. (2007) Lehtokari et al. (2007)	allelic to NEM4 (group 3), DA1A and DA2B (group 16)
Cap myopathy	3.43	AD		1q42.13	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Hung et al. (2010)	allelic to CMD with rigid spine (group 2), CFTD and NEM3 (group 3)
Congenital neuromuscular disease with uniform type 1 fiber (CNMDU1)	3.44	AR, AD	CCD 117000	19q13.2	***RYR1*** *180901*	Ryanodine receptor I	Sato et al. (2007)	allelic to CMD related to *RYR1* (group 2), CFTD, CNM related to *RYR1*, CCD, minicore myopathy with external ophthalmoplegia, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to *RYR1* (group 16)

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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)
Salih myopathy, (Congenital myopathy with fatal cardiomyopathy)	3.45	AR	SALMY 611705	2q31.2	***TTN*** *188840*	Titin	Carmignac et al. (2007)	Allelic to LGMDR10 (group 1), CNM related to *TTN* (group 3), MmD related to *TTN* (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A)
Congenital skeletal myopathy and fatal cardiomyopathy	3.46	AR		11p11.2	***MYBPC3*** *600958*	Cardiac myosin binding protein-C	Tajsharghi et al. (2010)	allelic to CMH4, CMD1MM and LVNC10 (group 10A)
Congenital myopathy Compton-North	3.47	AR	MYPCN 612540	12q12	***CNTN1*** *600016*	Contactin-1	Compton et al. (2008)	
Sarcotubular myopathy	3.48	AR		9q33.1	***TRIM32*** *602290*	Tripartite motif containing 32	Schoser et al. (2005)	allelic to LGMDR8 (group 1)
Congenital myopathy related to *HACD1*	3.49	AR		10p12.33	***HACD1*** *(PTPLA) 610467*	3-Hydroxyacyl-CoA dehydratase (Protein tyrosine phosphatase-like)	Muhammad et al. (2013)	allelic to ARVD6 (group 10B)
Congenital myopathy with ophthalmoplegia related to *CACNA1S*	3.50	AR		1q32.1	***CACNA1S*** *114208*	Calcium channel,voltage-dependent, L type, alpha 1S subunit	Hunter et al. (2015)	Allelic to HOKPP1 (group 7) MHS5 (group 8)
Congenital myopathy with neuropathy and deafness	3.51	AR	NEDHND 617519	19q13.2	***SPTBN4*** *606214*	Spectrin, beta, nonerythrocytic, 4	Knierim et al. (2017)	
Myopathy, congenital, with excess of muscle spindles	3.52	AD	CMEMS 218040	11p15.5	***HRAS*** *190020*	V-Ha-RAS Harvey rat sarcoma viral oncogene homolog	Quélin et al. (2017)	
Carey-Fineman-Ziter syndrome (formerly congenital myopathy with Moebius sequence and Robin sequence)	3.53	AR	CFZS 254940	9q34.2	***MYMK*** *615345*	Myomaker	Di Gioia et al. (2017)	
Myopathy, congenital, Baily-Bloch (Native American myopathy)	3.54	AR	MYPBB 255995	12q13.3	***STAC3*** *615521*	SH3 and cysteine-rich domains 3	Horstick et al. (2013)	Allelic to Myopathy, congenital, with malignant hyperthermia susceptibility (group 3)
Myopathy, congenital, with malignant hyperthermia susceptibility	3.55	AR		12q13.3	***STAC3*** *615521*	SH3 and cysteine-rich domains 3	Zaharieva et al. (2018)	Allelic to MYPBB (group 3)
Myopathy congenital with structured cores and Z-line abnormalities	3.56	AD	MYOCOZ 618654	1q43	***ACTN2*** *102573*	Actinin, alpha 2	Lornage et al. (2019)	Allelic to MPD6 (group 4), CMH23 and CMD1AA (group 10A)
Myopathy congenital with fast twitch (type II) fiber atrophy	3.57	AR	MYOFTA 618414	2q34	***MYL1*** *160780*	Myosin, light polypeptide 1, alkali, skeletal fast	Ravenscroft et al. (2018)	
Congenital amyotrophy	3.58	AR		16p13.3	***CACNA1H*** *607904*	Calcium channel, voltage-dependent, T type, aplpha-1H subunit	Carter et al. (2019)	
Congenital multi-minicore myopathy	3.59	AR		3q26.33	***FXR1*** *600819*	FMR1 autosomal homolog	Estan et al. (2019)	
Congenital Myopathy related to PAX7	3.60	AR		1p36.13	***PAX7*** *167410*	Paired Box gene 7	Feichtinger et al. (2019)	
Congenital Myopathy related to *SCN4A*	3.61	AR		17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Zaharieva et al. (2016) Gonorazky et al. (2017) Sloth et al. (2018)	allelic to HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to *SCN4A* (group 16)
Congenital Myopathy related to *PYROXD1*	3.62	AR		12p12.1	***PYROXD1*** *617220*	Pyridine nucleotide-disulphide oxidoreductase domain 1	Lornage et al. (2019)	allelic to LGMD related to *PYROXD1* (group 1) and MMF8 (group 5)
Congenital Myopathy related to *TNPO3*	3.63	AD		7q32.1	***TNPO3*** *610032*	Transportin 3	Vihola et al. (2019) Angelini et al. (2019)	allelic to LGMDD2 (group 1)
Scapuloperoneal myopathy, *MYH7*-related	3.64	AD	SPMM 181430	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Pegoraro et al. (2007)	Allelic to CFTD, MSMA, MSMB, eccentric core disease (group 3); MPD1 (group 4); CMH1, CMD1S and LVNC5 (group 10A)
Congenital myopathy related to *UNC45B*	3.65	AR		17q12	***UNC45B*** *611220*	UNC45 Myosin Chaperone B	Dafsari et al. (2019)	

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Dusty core disease related to RYR1	3.66	AR	DuCD	19q13.2	***RYR1*** *180901*	Ryanodine receptor I	Garibaldi et al. (2019)	allelic to CMD related to RYR1 (group 2), CFTD, CNM related to RYR1, CCD, minicore myopathy with external ophthalmoplegia, CNMDU1 (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to RYR1 (group 16)
Lysosomal storage myopathy	3.67	AR	ML4 252,650	19p13.2	***MCOLN1*** *605248*	Mucopilin 1	Zambon et al. (2021)	
Congenital myopathy	3.68	AD		20q13.12	***TNNC2*** *191039*	Troponin C Fast	van de Locht (2021)	
GROUP 4. DISTAL MYOPATHIES								
Miyoshi muscular dystrophy 1	4.1	AR	MMD1 254130	2p13.2	***DYSF*** *603009*	Dysferlin	Bejaoui et al. . (1995) Bashir et al. (1998) Liu et al. (1998)	allelic to LGMDR2 (group 1)
Tibial muscular dystrophy (Udd)	4.2	AD	TMD 600334	2q31.2	***TTN*** *188840*	Titin	Haravuori et al. (1998, 2001) Hackman et al. (2002)	allelic to LGMDR10 (group 1), CNM related to *TTN,* MmD related to *TTN* and SALMY (group 3), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A)
Nonaka Myopathy	4.3	AR	NM 605820	9p13.3	***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 1 (Laing)	4.4	AD	MPD1 160500	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Laing et al. (1995) Mastaglia et al. (2000) Meredith et al. (2004)	allelic to CFTD, MSMA, MSMB, SPMM, eccentric core disease (group 3), CMH1, CMD1S and LVNC5 (group 10A)
Vocal cord and pharyngeal distal myopathy (VCPDM) reclassfied as ALS21	4.5	AD	ALS21 606070	5q31.2	***MATR3*** *164015*	Matrin 3	Feit et al. (1998) Senderek et al. (2009)	Allelic to ALS21 (group 12)
Myopathy distal 3	4.6	AD	MPD3 610099	8p22-q11 12q13-q22	*?*	?	Haravuori et al. (2004)	
Welander distal myopathy	4.7	AD	WDM 604454	2p13.3	***TIA1*** *603518*	Cytotoxic granule-associated RNA binding protein	Ahlberg et al. (1999) Hackman et al. (2013) Klar et el (2013)	allelic to Welander-like syndrome (group 4)
Welander-like distal myopathy	4.8	Digenic		5q35.3 2p13.3	***SQSTM1*** *601530* + ***TIA1*** *603518*	Sequestosome 1 + Cytotoxic granule-associated RNA binding protein	Lee et al. (2018)	***SQSTM1*** Allelic to DMRV (group 4) and FTDALS3 (group 12) ***TIA1*** Allelic to WDM (group 4)
Vacuolar neuromyopathy	4.9	AD	601846	19p13.3	*?*	?	Servidei et al. (1999)	
Distal myopathy with myotilin defect	4.10	AD		5q31.2	***MYOT*** *604103*	Myotilin	Penisson-Besnier et al. (1998, 2006)	allelic to formerly LGMD1A (group 1), MFM3 (group 5), spheroid body myopathy (group 5)
Distal myopathy with nebulin defect	4.11	AR		2q23.3	***NEB*** *161650*	Nebulin	Wallgren-Pettersson et al. (2007)	allelic to NEM2 (group 3) and AD Distal myopathy with nebulin defect (group 4)
Distal myopathy with nebulin defect	4.12	AD		2q23.3	***NEB*** *161650*	Nebulin	Kiiski et al. (2019)	allelic to NEM2 (group 3) and AR Distal myopathy with nebulin defect (group 4)
Distal myopathy, Tateyama type	4.13	AD	MPDT 614321	3p25.3	***CAV3*** *601253*	Caveolin-3	Tateyama et al. (2002) Fulizio et al. (2005)	allelic to formerly LGMD1C (group 1), Creatin phosphokinase elevated serum (group 5), RMD2 (group 6), CMH1 (group 10A) and LQT9 (group 10B)
Late onset distal myopathy (Markesbery-Griggs)	4.14	AD		10q23.2	***LDB3*** *605906*	LIM domain binding-3	Griggs et al. (2007)	allelic to MFM4 (group 5), CMD1C, CMH24 and LVNC3 (group 10A)
Miyoshi muscular dystrophy 3	4.15	AR	MMD3 613319	11p14.3	***ANO5*** *608662*	Anoctamin 5	Bolduc et al. (2010)	allelic to LGMDR12 (group 1) and Muscular dystrophy with gnathodiaphyseal dysplasia (group 1)

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Distal myopathy related to *DNM2*	4.16	AD		19p13.2	***DNM2*** *602378*	Dynamin 2	Fischer et al. (2006)	allelic to CMD related to *DNM2*(group 2), CNM1 (group 3), LCCS5 (group 12) and CMTDIB (group 14)
Early onset distal myopathy with *KLHL9* defect	4.17	AD		9p21.3	***KLHL9*** *611201*	Kelch-like 9	Cirak et al. (2010)	
Distal Myopathy 4	4.18	AD	MPD4 614065	7q32.1	***FLNC*** *102565*	Filamin C (gamma)	Duff et al. (2011)	allelic to MFM5 (group 5), CMH26 and RCM5 (group 10A)
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 1	4.19	AD	IBMPFD1 167320	9p13.3	***VCP*** *601023*	Valosin-containing protein	Palmio et al. (2011)	allelic to Scapuloperoneal myopathy (group 1), IBMPFD1 (group 5), ALS14 (group 12) and CMT2Y (group 14)
Distal myopathy 5	4.20	AR	MPD5 617030	14q32.33	***ADSSL1*** *612498*	Adenylosuccinate synthase-like	Park et al. (2016)	
Myopathy, distal, with rimmed vacuoles	4.21	AD	DMRV 617158	5q35.3	***SQSTM1*** *601530*	Sequestosome 1	Bucelli et al. (2015)	allelic to Welander-like syndrome (group 4) and FTDALS3 (group 12)
Myopathy, distal, with rimmed vacuoles	4.22	AD		7q36.3	***DNAJB6*** *611332*	Hsp40 homologue, subfamily B, number 6	Ruggieri et al. (2015)	allelic to LGMDD1 (group 1) and distal myopathy (group 4)
Distal myopathy	4.23	AD		7q36.3	***DNAJB6*** *611332*	Hsp40 homologue, subfamily B, number 6	Palmio et al. (2019)	Allelic to LGMDD1 (group 1), Rimmed vacuole distal myopathy (group 4)
Rimmed vacuole myopathy	4.24	AD		12q24.23	***HSPB8*** *608014*	Heat-shock 22 kD protein 8	Al-Tahan et al. (2019)	Allelic to distal myopathy and motor neuropathy (group 4) HMN2A (group 12) and CMT2L (group 14)
Distal Myopathy 6, Adult-onset	4.25	AD	MPD6 618655	1q43	***ACTN2*** *102573*	Actinin alpha-2	Savarese et al. (2019)	Allelic to MYOCOZ (group 3), CMH23 and CMD1AA (group 10A)
Distal Myopathy and motor neuropathy	4.26	AD		12q24.23	***HSPB8*** *608014*	Heat-shock 22 kD protein 8	Ghaoui et al. (2016)	Allelic to Rimmed vacuole myopathy (group 4), HMN2A (group 12) and CMT2L (group 14)
Distal Myopathy with protein inclusions	4.27	XLD		Xp22.12	***SMPX*** *300226*	Small Muscle Protein, X-linked	Johari et al. (2021)	
Distal Myopathy	4.28	AD		19p13.3	***PLIN4*** *613247*	Perilipin 4	Ruggieri et al. (2020)	
GROUP 5. OTHER MYOPATHIES								
Myofibrillar myopathies								
Myofibrillar myopathy 1	5.1	AD	MFM1 601419	2q35	***DES*** *125660*	Desmin	Goldfarb et al. (1998) Munoz-Marmol et al. (1998)	allelic to formerly LGMD1 related to *DES* and formerly LGM2R (group1), MFM1 with ARCV7 (group 5), CMD1I (group 10A) and ARVC7 (group 10B)
Myofibrillar myopathy 2	5.2	AD	MFM2 608810	11q23.1	***CRYAB*** *123590*	Alpha-B crystallin	Vicart et al. (1998) Selcen et al. (2003)	allelic to CMD1II (group 10A)
Myofibrillar myopathy 3	5.3	AD	MFM3 609200	5q31.2	***MYOT*** *604103*	Myotilin	Selcen and Engel (2004)	allelic to formerly LGMD1A (group 1), distal myotilinopathy (group 4), spheroid body myopathy (group 5)
Spheroid body myopathy	5.4	AD	182,920	5q31.2	***MYOT*** *604103*	Myotilin	Foroud et al. (2005)	allelic to formerly LGMD1A (group 1), distal myotilinopathy (group 4), MFM3 (group 5)
Myofibrillar myopathy 4	5.5	AD	MFM4 609452	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Selcen and Engel (2005)	allelic to Late onset distal myopathy (group 4), CMD1C, CMH24 and LVNC3 (group 10A)
Myofibrillar myopathy 5	5.6	AD, AR	MFM5 609524	7q32.1	***FLNC*** *102565*	Filamin C (gamma)	Vorgerd et al. (2005) Kölbel et al. (2020) Schuld et al. (2020)	allelic to MPD4 (group 4), CMH26 and RCM5 (group 10A)
Myofibrillar myopathy 6	5.7	AD	MFM6 612954	10q26.11	***BAG3*** *603883*	BCL2-associated athanogene 3	Selcen et al. (2009)	allelic to CMD1HH (group 10A) and CMT related to BAG3 (group 14)
Myofibrillar myopathy 7	5.8	AR	MFM7 617114	3q22.2	***KY*** *605739*	Kyphoscoliosis peptidase	Hedberg-Oldfors et al. (2016) Straussberg et al. (2016)	

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Myofibrillar myopathy 8	5.9	AR	MFM8 617258	12p12.1	***PYROXD1*** *617220*	Pyridine nucleotide-disulphide oxidoreductase domain 1	O'Grady et al. (2016)	allelic to LGMD related to *PYROXD1* (group 1) and Congenital Myopathy related to *PYROXD1* (group 3)
Myofibrillar myopathy 9 with early respiratory failure	5.10	AD	MFM9 (HMERF) 603689	2q31.2	***TTN*** *188840*	Titin	Nicolao et al. (1999) Lange et al. (2005)	allelic to LGMDR10 (group 1), CMN related to *TTN* (group3), MmD related to *TTN* (group 3), SALMY (group 3), TMD (group 4), CMH9 (group 10A), CMD1G (group 10A), LCCS related to TTN (group 12)
Myofibrillar myopathy 10	5.11	AR	MFM10 619040	10p11.23	***SVIL*** *604126*	Supervillin	Hedberg-Oldfors et al. (2020)	
Desmin-related myopathy with Mallory bodies	5.12	AD	RSMD1 602771	1p36.11	***SELENON*** *(formerly SEPN1) 606210*	Selenoprotein N	Ferreiro et al. (2004)	allelic to RSMD1 (group 2), CFTD (group 3) Multiminicore disease (group 3)
Cardiac and skeletal aggregate myopathy	5.13	Digenic		1p36.11 2p.23.3	***TRIM63*** *06,131* + ***TRIM54*** *606474*	Tripartite motif containing 63 (MURF1) + Ring finger protein 30 (MURF3)	Olivé et al. (2015)	
Myofibrillar myopathy with arrhythmogenic right ventricular cardiomyopathy (ARCV7) also listed in group 10B	5.14	AD	MFM1 601419	10q22 > 2q35	***DES*** *125660*	Desmin	Melberg et al. (1999) Kuhl et al. (2008) Hedberg et al. (2012)	allelic to formerly LGMD1 related to *DES* and formerly LGM2R (group 1), MFM1 (group 5), CMD1I (group 10A) and ARVC7 (group 10B)
Miscellaneous								
Danon disease	5.15	XD	GSD2B 300257	Xq24	***LAMP2*** *309060*	Lysosomal-associated membrane protein 2	Nishino et al. (2000) Musumeci et al. (2005)	
Myopathy with excessive autophagy	5.16	XR	MEAX 310440	Xq28	***VMA21*** *300913*	S. Cerevisiae homolog of VMA21	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	5.17	AR	CLN3 204200	16p12.1	***CLN3*** *607042*	Ceroid-lipofuscinosis, neuronal 3 (= Battenin)	Cortese et al. (2014)	
Oculopharyngeal muscular dystrophy	5.18	AD	OPMD 164300	14q11.2	***PABPN1*** *602279*	Polyadenylate-binding protein, nuclear 1	Brais et al. (1995, 1998) Robinson et al. (2005)	
Oculopharyngodistal myopathy 1	5.19	AD	OPDM1 164310	8q22.3	***LRP12*** *618299*	Low density lipoprotein receptor-related protein 12	Ishiura et al. (2019)	
Oculopharyngodistal myopathy 2	5.20	AD	OPDM2 618940	19p13.12	***GIPC1*** *605072*	GIPC PDZ Domain-containing family, member 1	Deng et al. (2020)	
Epidermolysis bullosa simplex associated with muscular dystrophy	5.21	AR	EBSMD 226670	8q24.3	***PLEC*** *601282*	Plectin	Gache et al. (1996) Smith et al. (1996) Wuyts et al. (1996)	allelic to LGMDR17 (group 1), LGMD with ophthalmoplegia (group 1), myasthenic syndrome related to *PLEC* (group 11)
Muscle hypertrophy	5.22	AR	MSLHP 614160	2q32.2	***MSTN*** *601,788*	Myostatin	Schuelke et al. (2004)	
Fibrodysplasia ossificans progressiva	5.23	AD	FOP 135100	2q24.1	***ACVR1*** *102576*	Activin A receptor, type 1	Shore et al. (2006)	
Creatine phosphokinase, elevated serum (formerly HyperCKemia, iodiopathic)	5.24	AD	123320	3p25.3	***CAV3*** *601253*	Caveolin-3	Carbone et al. (2000)	allelic to formerly LGMD1C (group 1), MPDT (group 4), RMD2 (group 6), CMH1 and LQT9 (group 10)
X-linked myopathy with postural muscle atrophy	5.25	XR	XMPMA 300696	Xq26.3	***FHL1*** *300163*	Four-and-a-half LIM domains 1	Windpassinger et al., 2008	allelic to EDMD6 (group 1), RBMX1A/B (group 5), SPM (group 5)
Scapuloperoneal myopathy	5.26	XD	SPM 300695	Xq26.3	***FHL1*** *300163*	Four-and-a-half LIM domains 1	Quinzii et al. (2008)	allelic to EDMD6 (group 1), RBMX1A/B (group 5), XMPMA (group 5)
Reducing body myopathy (1A and 1B)	5.27	XD	RBMX1A 300717 RBMX1B 300718	Xq26.3	***FHL1*** *300163*	Four-and-a-half LIM domains 1	Schessl et al. (2008), Shalaby et al. (2009)	allelic to EDMD6 (group 1), XMPMA (group 5), SPM (group 5)

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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 muscle weakness, X-linked	5.28	XR	EMWX 300211	Xp22.3	*?*	?	Ryan et al. (1999)	
Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia 1	5.29	AD	IBMPFD1 167320	9p13.3	***VCP*** *601023*	Valosin-containing protein	Watts et al. (2004) Haubenberger et al. (2005)	allelic to scapuloperoneal myopathy (group 1), IBMPFD1 (group 4), ALS14 (group 12) and CMT2Y (group 14)
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2	5.30		IBMPFD2 615422	7p15.2	***HNRNPA2B1*** *600124*	Heterogeneous nuclear ribonucleoprotein A2/B1	Kim et al. (2013)	
Myopathy with lactic acidosis, hereditary	5.31	AR	HML 255125	12q23.3	***ISCU*** *611911*	Iron-sulfur cluster scaffold homolog (E. coli)	Mochel et al. (2008)	
Late onset axial myopathy related to *RYR1*	5.32	AD		19q13.2	***RYR1*** *180901*	Ryanodine receptor 1 (skeletal)	Løseth et al. (2013)	allelic to CMD related to *RYR1* (group 2), CFTD, CNM related to *RYR1*, CCD, CNMDU1, minicore myopathy with external ophthalmoplegia, DuCD (group 3), MHS1 (group 8), Fetal akinesia related to RYR1 (group 16)
Tubular aggregate myopathy 1	5.33	AD	TAM1 160565	11p15.4	***STIM1*** *605921*	Stromal interaction molecule 1	Bohm et al. (2013) Hedberg et al. (2014) Nesin et al. (2014)	Allelic to muscle weakness with hyperlaxity, orofacial abnormalities and respiratory infections (group 5)
Tubular aggregate myopathy 2	5.34	AD	TAM2 615883	12q24.31	***ORAI1*** *610277*	ORAI calcium release-activated calcium modulator 1	Nesin et al. (2014)	
Vacuolar myopathy with CASQ1 aggregates	5.35	AD	VMCQA 616231	1q23.2	***CASQ1*** *114250*	Calsequestrin 1	Rossi et al. (2014)	
Myopathy with characteristic sarcoplasmic inclusions	5.36	AD		22q12.3	***MB*** *160000*	Myoglobin	Olivé et al. (2019)	
Myopathy with extrapyramidal signs	5.37	AR	MPXPS 615673	10q22.1	***MICU1*** *605084*	Mitochondrial Calcium Uptake Protein 1	Logan et al. (2014)	
Oculopharyngo-distal myopathy	5.38	AD		1q21.2	***NOTCH2NLC*** *(NBPF19)* *618025*	Notch2 N-terminal-like protein	Ogasawara et al. (2020) Yu et al. (2021)	Allelic to Neuronal intranuclear inclusion diseases (group 14)
Muscle weakness with hyperlaxity, orofacial abnormalities and respiratory infections	5.39	AR		11p15.4	***STIM1*** *605921*	Stromal interaction molecule 1	Salvi et al. (2021)	Allelic to TAM1 (group 5)
GROUP 6. MYOTONIC SYNDROMES								
Myotonic dystrophy 1 (Steinert)	6.1	AD	DM1 160900	19q13.32	***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	3q21.3	***CNBP*** *(formerly ZNF9)* *116955*	CCHC-type Zinc finger nucleic acid-binding protein (Zinc finger protein 9)	Ranum et al. (1998) Liquori et al. (2001)	
Myotonia, dominant (Thomsen)	6.3	AD	see under Ion channel muscle diseases (group 7)					
Myotonia, recessive (Becker)	6.4	AR	see under Ion channel muscle diseases (group 7)					
Rippling muscle disease, dominant	6.5	AD	RMD1 600332	1q41	*?*	?	Stephan et al. (1994)	
Rippling muscle disease, dominant	6.6	AD	RMD2 606072	3p25.3	***CAV3*** *601253*	Caveolin-3	Betz et al. (2001)	allelic to formerly LGMD1C (group 1), MPDT (group 4), Creatin phosphokinase elevated serum (group 5), CMH1 (group 10A) and LQT9 (group 10B)

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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)
Rippling muscle disease, recessive	6.7	AR	RMD2 606072	3p25.3	***CAV3*** *601253*	Caveolin-3	Kubisch et al. (2003, 2005)	allelic to formerly LGMD1C (group 1), MPDT (group 4), Creatin phosphokinase elevated serum (group 5), CMH1 (group 10A) and LQT9 (group 10B)
Schwartz-Jampel syndrome	6.8	AR	SJS1 255800	1p36.12	***HSPG2*** *14246*	Heparan sulfate proteoglycan of basement membrane (perlecan)	Nicole et al. (1995, 2000)	
Brody disease	6.9	AR	601003	16p11.2	***ATP2A1*** *(formerly SERCA1) 108730*	ATPase, Ca^{++}transporting, fast twitch 1	Odermatt et al. (1996)	
Group 7. ION CHANNEL MUSCLE DISEASES								
Chloride channel								
Myotonia congenita, dominant (Thomsen)	7.1	AD	THD 160800	7q34	***CLCN1*** *118425*	Muscle chloride channel	Koch et al. (1992) George et al. (1993)	allelic to Becker myotonia (group 7)
Myotonia, recessive (Becker)	7.2	AR	255700	7q34	***CLCN1*** *118425*	Muscle chloride channel	Koch et al. (1992)	allelic to Thomsen myotonia (group 7)
Sodium channel								
Hyperkalaemic periodic paralysis	7.3	AD	HYPP 170500	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Fontaine et al. (1990) Ptáček et al. (1991) Rojas et al. (1991) Miller et al. (2004)	allelic to Congenital Myopathy related to *SCN4A* (group 3), HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to *SCN4A* (group 16)
Hypokalaemic periodic Paralysis, type 2	7.4	AD	HOKPP2 613345	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Bulman et al. (1999) Jurkat-Rott et al. (2000)	allelic to Congenital Myopathy related to *SCN4A* (group 3), HYPP, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to *SCN4A* (group 16)
Hypokalaemic periodic paralysis	7.5	AD		1q23.2	***ATP1A2*** *182340*	ATPase, NA+/*K*+ transporting alpha-2 polypeptide	Sampedro et al. (2018)	
Paramyotonia congenita	7.6	AD	PMC 168300	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Ptáček et al. (1991–1993) Ebers et al. (1991) Koch et al. (1992) Mc Clatchey et al. (1992)	allelic to Congenital Myopathy related to *SCN4A* (group 3), HYPP, HOKPP2 and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to *SCN4A* (group 16)
Potassium-aggravated myotonia	7.7	AD	608390	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Ptáček et al. (1992, 1994) Heine et al. (1993) Lerche et al. (1993)	allelic to Congenital Myopathy related to *SCN4A* (group 3), HYPP, HOKPP2 and PMC (group 7), CMS16 (group 11), Severe foetal hypokinesia related to *SCN4A* (group 16)
Calcium channel								
Hypokalaemic periodic paralysis, type 1	7.8	AD	HOKPP1 170400	1q32.1	***CACNA1S*** *114208*	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Fontaine et al. (1994) Ptáček et al. (1994) Jurkat-Rott et al. (1994) Elbaz et al. (1995)	allelic to congenital myopathy with ophthalmoplegia (group 3), MHS5 (group 8)
Acetazolamide responsive hereditary paroxysmal cerebellar ataxia (also listed in group 13 "Ataxias")	7.9	AD	APCA 108500	19p13.13	***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), APCA and EA2 and familial hemiplegic migraine (group 13)
Episodic ataxia type-2	7.10	AD	EA2 108500	19p13.13	***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 and 13), EA2 and familial hemiplegic migraine (group 13), SCA6 (group 13)
Potassium channel								
Hypokalaemic periodic paralysis	7.11	AD	HOKPP 170400	11q13.4	***KCNE3*** *604433*	Potassium channel, voltage-gated, Isk-related family, member 3	Abbott et al. (2001)	Allelic to BRGDA6 (group 10B)

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Episodic ataxia/myokymia syndrome	7.12	AD	EA1 160120	12p13.32	***KCNA1*** *176260*	Potassium channel, voltage-gated, shaker-related subfamily, member 1	Browne et al. (1994) Adelman et al. (1995)	
Thyrotoxic hypokalemic periodic paralysis	7.13		TTPP2 613239	17p11.2	***KCNJ18*** *613236*	Potassium channel, inwardly rectifying, subfamily J, member 18 (Kir2.6)	Ryan et al. (2010)	
Periodic paralysis, potassium sensitive cardiodysrythmic (Andersen's syndrome)		see LQ7 under hereditary cardiomyopathies (group 10B, online only)						
Long QT syndromes		see under hereditary cardiomyopathies (group 10B, online only)						
GROUP 8. MALIGNANT HYPERTHERMIAS								
Malignant hyperthermia	8.1	AD	MHS1 145600	19q13.2	***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 CMD related to *RYR1* (group 2) CFTD, CNM related to *RYR1*, CCD, CNMDU1, minicore myopathy with external ophthalmoplegia, DuCD (group 3), late onset axial myopathy (group 5), Fetal akinesia related to *RYR1* (group 16)
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	1q32.1	***CACNA1S*** *114208*	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Monnier et al. (1997)	allelic to congenital myopathy with ophthalmoplegia (group 3), HOKPP1 (group 7)
Malignant hyperthermia	8.6	AD	MHS6 601888	5p	*?*	?	Robinson et al. (1997)	
GROUP 9. METABOLIC MYOPATHIES								
Glycogen storage diseases								
Glycogen storage disease type II, Pompe disease (*also listed in group 10A)*	9.1	AR	GSD2 232300	17q25.3	***GAA*** *606800*	Glucosidase, alpha acid	Hers et al. (1963) Martiniuk et al. (1990) Wokke et al. (1995)	Allelic to Pompe Disease (formerly LMGD2V) (group 1)
Glycogen storage disease type III	9.2	AR	GSD3 232400	1p21.2	***AGL*** *610860*	Amylo-1, 6-glucosidase, 4-alpha-glucano transferase (glycogen debrancher enzyme)	Sheng et al. (1996)	
Glycogen storage disease type IV	9.3	AR	GSD4 232500	3p12.2	***GBE1*** *607839*	Glycogen branching enzyme (1,4-α-glucan branching enzyme)	Brown et al. (1966) Bao et al. (1996) Bruno et al. (2004)	
Glycogen storage disease type V (McArdle)	9.4	AR	GSD5 232600	11q13.1	***PYGM*** *608455*	Glycogen phosphorylase, muscle	Mommaerts et al. (1959) Schmidt et al. (1959) Lebo et al. (1984) Tsujino et al. (1993)	
Glycogen storage type VII (Tarui)	9.5	AR	GSD7 232800	12q13.11	***PFKM*** *610681*	Phosphofructokinase, muscle type	Tarui et al. (1965) Nakajima et al. (1991) Howard et al. (1996)	
Glycogen storage disease type IXd (ex type VIII)	9.6	XR	GSD9D 300559	Xq13.1	***PHKA1*** *311870*	Phosphorylase kinase, alpha-1 subunit	Wehner et al. (1994) Burwinkel et al. (2004)	

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Glycogen storage disease type XIV (Congenital disorder of glycosylation, type It)	9.7	AR	CDG1T 614921	1p31.3	***PGM1*** *171900*	Phosphoglucomutase 1	Stojkovic et al. (2009)	
Glycogen storage disease type XV	9.8	AR	GSD15 613507	3q24	***GYG1*** *603942*	Glycogenin 1	Moslemi et al. (2010)	allelic to PGBM2 (group 9)
Glycogen storage disease type 0	9.9	AR	GSD0B 611556	9q13.33	***GYS1*** *138570*	Glycogen synthase 1	Kolberg et al. (2007)	
Glycogen storage disease of heart, lethal congenital	9.10	AD	261740	7q36.1	***PRKAG2*** *602743*	Protein kinase, AMP-activated, non catalytic (AMPK-gamma-2)	Burwinkel et al. (2005)	allelic to CMH6 (group 10A)
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	Nilsson et al. (2013)	
Polyglucosan Body Myopathy 2	9.12	AR	PGBM2 616199	3q24	***GYG1*** *603942*	Glycogenin 1	Malfatti et al. (2014)	allelic to GSD15 (group 9)
Glycolytic pathway								
Posphoglycerate kinase 1 deficiency	9.13	XR	300653	Xq21.1	***PGK1*** *311800*	Phosphoglycerate kinase 1	DiMauro et al. (1981, 1983) Rosa et al. (1982)	
Glycogen storage disease type X	9.14	AR	GSD10 261670	7p13	***PGAM2*** *612931*	Phosphoglycerate mutase 2	DiMauro et al. (1981) Edwards et al. (1989) Castella-Escola et al. (1990) Tsujino et al. (1993)	
Glycogen storage disease type XI	9.15	AR	GSD11 612933	11p15.1	***LDHA*** *150000*	Lactate dehydrogenase A	Kanno et al. (1980) Scrable et al. (1990)	
Glycogen storage disease type XIII	9.16	AD	GSD13 612932	17p13.2	***ENO3*** *131370*	Enolase 3	Comi et al. (2001)	
Erythrocyte lactate transporter defect (Myopathy due to actate transporter defect)	9.17	AD	245340	1p13.2	***SLC16A1*** *600682*	Solute carrier family 16 (monocarboxylic acid transporter), member 1	Merezhinskaya et al. (2000)	
Disorders of lipid metabolism								
Carnitine palmitoyl-transferase II deficiency, myopathic, stress induced	9.18	AR	255110	1p32.3	***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	9.19	AR	CDSP 212140	5q31.1	***SLC22A5*** *603377*	Solute carrier family 22 (organic cation transporter), member 5	Nezu et al. (1999)	
Carnitine/acyl-carnitine translocase deficiency	9.20	AR	CACTD 212138	3p21.31	***SLC25A20*** *613698*	Solute carrier family 25 (carnitine/acylcarnitine translocase), member 20	Huizing et al. (1997) Ogawa et al. (2000)	
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIA)	9.21	AR	MADD 231680	15q24.2-q24.3	***ETFA*** *608053*	Electron-transfer-flavoprotein, alpha polypeptide	Indo et al. (1991) Freneaux et al. (1992)	
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIB)	9.22	AR	MADD 231680	19q13.41	***ETFB*** *130410*	Electron-transfer-flavoprotein, beta polypeptide	Colombo et al. (1994)	
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIC, riboflavin responsive)	9.23	AR	MADD 231680	4q32.1	***ETFDH*** *231675*	Electron-transfer-flavoprotein dehydrogenase	Beard et al. (1993)	
Acyl-CoA dehydrogenase (very long chain) deficiency (VLCAD deficiency)	9.24	AR	ACADVLD 201475	17p13.1	***ACADVL*** *609575*	Acyl-Coenzyme A dehydrogenase, very long chain	Aoyama (1993, 1995) Strauss et al. (1995) Mathur et al. 1999)	
Mitochondrial complex I deficiency due to ACAD9 deficiency	9.25	AR	611126	3q21.3	***ACAD9*** *611103*	Acyl-CoA dehydrogenase family member 9 (**M**)	Fragaki etal (2016)	

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Triglyceride storage disease with ichthyosis (Chanarin-Dorfman syndrome)	9.26	AR	CDS 275630	3p21.33	***ABHD5*** *604780*	Abhydrolase domain containing 5	Lefevre et al. (2001)	
Neutral lipid storage disease with myopathy without ichthyosis	9.27	AR	NLSDM 610717	11p15.5	***PNPLA2*** *609059*	Patatin-like phosphorylase domain-containing protein 2 (Adipose triglyceride lipase)	Fischer et al. (2007)	
Acute Recurrent myoglobinuria	9.28	AR	268200	2p25.1	***LPIN1*** *605518*	lipin 1 (phsosphatidic acid phosphatase 1)	Zeharia et al. (2008)	
Mitochondrial myopathy with lactic acidosis	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 10A. HEREDITARY CARDIOMYOPATHIES non arrhythmogenic								
Hypertrophic cardiomyopathies								
Familial hypertrophic cardiomyopathy, 1	10.1	AD	CMH1 192600	14q11.2	***MYH7*** *160760*	Myosin heavy chain 7 (beta), cardiac muscle	Jarcho et al. (1989) Solomon et al. (1990) Tanigawa et al. (1990) Geisterfer-Lowrance et al. (1990)	allelic to CFTD, MSMA, MSMB, SPMM, eccentric core disease (group 3), MPD1 (group 4), CMD1S and LVNC5 (group 10A)
Familial hypertrophic cardiomyopathy, 2	10.2	AD	CMH2 115195	1q32.1	***TNNT2*** *191045*	Cardiac troponin T	Watkins et al. (1993) Thierfelder et al. (1994)	allelic to CMD1D, RCM3 and LVNC6 (group 10A)
Familial hypertrophic cardiomyopathy, 3	10.3	AD	CMH3 115196	15q22.2	***TPM1*** *191010*	Tropomyosin-1	Thierfelder et al. (1994)	allelic to CMD1Y and LVNC9 (group 10A)
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), CMD1MM and LVNC10 (group 10A)
Familial hypertrophic cardiomyopathy, 6	10.5	AD	CMH6 600858	7q36.1	***PRKAG2*** *602743*	Protein kinase, AMP-activated, non catalytic (AMPK-gamma-2)	Blair et al. (2001)	allelic to glycogen storage disease of heart, lethal congenital (group 9)
Familial hypertrophic cardiomyopathy, 7	10.6	AD	CMH7 613690	19q13.42	***TNNI3*** *191044*	Cardiac troponin I	Kimura et al. (1997)	allelic to RCM1, CMD1FF and CMD2A (group 10A)
Familial hypertrophic cardiomyopathy, 8	10.7	AD	CMH8 608751	3p21.31	***MYL3*** *160790*	Myosin, light chain 3, alkali; ventricular, skeletal, slow	Poetter et al. (1996)	
Familial hypertrophic cardiomyopathy, 9	10.8	AD	CMH9 613765	2q31.2	***TTN*** *188840*	Titin	Satoh et al. (1999)	allelic to LGMDR10 (group 1), CNM related to *TTN* (group3), MmD related to *TTN* (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMD1G (group 10A), LCCS related to TTN (group 12)
Familial hypertrophic cardiomyopathy, 10	10.9	AD	CMH10 608758	12q24.11	***MYL2*** *160781*	Myosin, light chain 2, regulatory, cardiac, slow	Poetter et al. (1996)	Allelic to CFTD (group 3)
Familial hypertrophic cardiomyopathy, 11	10.10	AD	CMH11 612098	15q14	***ACTC1*** *102540*	Actin, alpha, cardiac muscle	Mogensen et al. (1999)	alllelic to CMD1R, LVNC4 (group 10A)
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 10A)
Familial hypertrophic cardiomyopathy, 13	10.12	AD	CMH13 613243	3p21.1	***TNNC1*** *191040*	Slow troponin C	Landstrom et al. (2008)	allelic to CMD1Z (group 10A)
Familial hypertrophic cardiomyopathy, 14	10.13	AD	CMH14 613251	14q11.2	***MYH6*** *160710*	Myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al. (2005)	allelic to CMD1EE (group 10A) and SSS3 (group 10B)

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Hypertrophic cardiomyopathy, 15	10.14	AD	CMH15 613255	10q22.2	***VCL*** *193065*	Vinculin	Vasile et al. (2006)	allelic to CMD1W (group 10)
Familial hypertrophic cardiomyopathy, 1	10.15	AD, digenic	CMH1 192600	20q11.21	***MYLK2*** *606566*	Myosin light chain kinase 2	Davis et al. (2001)	
Familial hypertrophic cardiomyopathy, 1	10.16	AD	CMH1 192600	3p25.3	***CAV3*** *601253*	Caveolin-3	Hayashi et al. (2004) Fulizio et al. (2005)	allelic to formerly LGMD1C (group 1), Creatin phosphokinase elevated serum (group 5), MPDT (group 4), RMD2 (group 6) and LQT9 (group 10B)
Hypertrophic cardiomyopathy, 16	10.17	AD	CMH16 613838	4q26	***MYOZ2*** *605602*	Myozenin 2 (calsarcin 1)	Osio et al. (2007)	
Hypertrophic cardiomyopathy, 17	10.18	AD	CMH17 613873	20q13.12	***JPH2*** *605267*	Junctophilin-2	Landstrom et al. (2007) Matsuhita et al. (2007)	
Hypertrophic cardiomyopathy, 18	10.19	AD	CMH18 613874	6q22.31	***PLN*** *172405*	Phospholamban	Minamisawa et al. (2003) Landstrom et al. (2011)	allelic to CMD1P (group 10A)
Hypertrophic cardiomyopathy, 19	10.20	AD	CMH19 61387	19p13.11	***CALR3*** *611414*	Calreticulin 3	Chiu et al. (2007)	
Hypertrophic cardiomyopathy, 20	10.21	AD	CMH20 613876	1p31.1	***NEXN*** *613121*	Nexilin F-actin binding protein	Wang et al. (2010)	allelic to CMD1CC (group 10A)
Hypertrophic cardiomyopathy related to cardiac ankyrin repeat domain protein	10.22	AD		10q23.31	***ANKRD1*** *609599*	Ankyrin repeat domain-protein 1	Arimura et al. (2009)	allelic to dilated cardiomyopathy (group 10A)
Hypertrophic cardiomyopathy, 22	10.23	AD	CMH22 615248	10q21.3	***MYPN*** *608517*	Myopalladin	Purevjav et al. (2012)	allelic to NEM11 (group 3), CMD1KK and RCM4 (group 10A)
Hypertrophic cardiomyopathy, 23	10.24	AD	CMH23 612158	1q43	***ACTN2*** *102573*	Actinin alpha-2	Chiu C et al. (2010)	allelic to MYOCOZ (group 3), MPD6, distal myopathy with facial weakness (group 4), CMD1AA (group 10A)
Hypertrophic cardiomyopathy, 24	10.25	AD	CMH24 601493	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Theis et al. (2006)	allelic to Late onset distal myopathy (group 4), MFM4 (group 5), CMD1C and LVNC3 (group 10)
Hypertrophic cardiomyopathy, 25	10.26	AD	CMH25 607487	17q12	***TCAP*** *604488*	Titin-cap (telethonin)	Hayashi et al. (2004)	allelic to LGMDR7 (group 1), CMD related to *TCAP* (group 2), CMD1N (group 10A)
Hypertrophic cardiomyopathy, 26	10.27	AD	CMH26 617047	7q32.1	***FLNC*** *102565*	Filamin C	Valdes-Mas et al. (2014)	allelic to MFM5 (group 5) and MPD4 (group 4) and RCM5 (group 10A)
Hypertrophic cardiomyopathy, 27	10.28	AR	CMH27 618052	15q25.3	***ALPK3*** *617608*	Alpha Kinase 3	Almomani et al. (2016)	
Mitochondrial complex 1 deficiency, nuclear type 11	10.29	AR	MC1DN11 618234	15q15.1	***NDUFAF1*** *606934*	NADH dehydrogenase (ubiquinone) complex I, Assembly factor 1 (**M**)	Fassone et al. (2011)	
Combined oxydative phosphorylation deficiency 3	10.30	AR	COXPD3 610505	12q14.1	***TSFM*** *604723*	Ts translation elongation factor, mitochondrial (**M**)	Smeitink et al.(2006)	
Combined oxydative phosphorylation deficiency 8	10.31	AR	COXPD8 614096	6p21.1	***AARS2*** *612035*	Alanyl-tRNA synthetase 2, mitochondrial (**M**)	Götz et al. (2011)	
Combined oxydative phosphorylation deficiency 9	10.32	AR	COXPD9 614582	3q21.1	***MRPL3*** *607118*	Mitochondrial ribosomal protein L3 (**M**)	Galmiche et al. (2011)	
Combined oxydative phosphorylation deficiency 10	10.33	AR	COXPD10 614702	6q13	***MTO1*** *614667*	Mitochondrial tRNA translation optimization 1 (**M**)	Ghezzi et al. (2012) Baruffini et al. (2013)	
Combined oxydative phosphorylation deficiency 16	10.34	AR	COXPD16 615395	2q36.1	***MRPL44*** *611849*	Mitochondrial ribosomal protein L44 (**M**)	Carroll et al. (2013)	
Cardio-encephalo-myopathy, fatal infantile, due to cytochrome C oxidase deficiency 1	10.35	AR	CEMCOX1 604377	22q13.33	***SCO2*** *604272*	Cytochrome c oxidase assembly protein (**M**)	Papadopoulou et al. (1999)	allelic to CMT2 related to SCO2 (group 14)
Cardio-encephalo-myopathy, fatal infantile, due to cytochrome C oxidase deficiency 2	10.36	AR	CEMCOX2 615119	10q24.2	***COX15*** *603646*	Cytochrome c oxidase assembly factor Cox15 (**M**)	Antonicka et al. (2003)	

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Dilated cardiomyopathies								
Dilated cardiomyopathy, 1A	10.37	AD	CMD1A 115200	1q22	***LMNA*** *150330*	Lamin A/C	Fatkin et al. (1999)	allelic to EDMD2, EDMD3 and formerly LGMD1B (group1), MDCL (group 2), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
Dilated cardiomyopathy, 1B	10.38	AD	CMD1B 600884	9q13	*?*	?	Krajinovic et al. (1995)	
Dilated cardiomyopathy, 1C, with or without LVNC	10.39	AD	CMD1C 601493	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Bowles et al. (1996) Vatta et al. (2003) Arimura et al. (2004)	allelic to Late onset distal myopathy (group 4), MFM4 (group 5), CMH24 and LVNC3 (group 10A)
Dilated cardiomyopathy, 1D	10.40	AD	CMD1D 601494	1q32.1	***TNNT2*** *191045*	Troponin type T2 (cardiac)	Durand et al. (1995) Kamisago et al. (2000)	allelic to CMH2, RCM3 and LVNC6 (group 10A)
Dilated cardiomyopathy, 1E	10.41	AD	CMD1E 601154	3p22.2	***SCN5A*** *600163*	Sodium channel, voltage-gated, type V, alpha	McNair et al. (2004)	allelic to BRGDA1, ATFB10, LQT3, SSS1 (group 10B)
Dilated cardiomyopathy, 1G	10.42	AD	CMD1G 604145	2q31.2	***TTN*** *188840*	Titin	Siu et al. (1999) Gerull et al. (2002) Itoh-Satoh et al. (2002)	allelic to LGMDR10 (group 1), CNM related to *TTN* (group 3), MmD related to *TTN* (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), LCCS related to TTN (group 12)
Dilated cardiomyopathy, 1H	10.43	AD	CMD1H 604288	2q14-q22	*?*	?	Jung et al. (1999)	
Dilated cardiomyopathy, 1I	10.44	AD	CMD1I 604765	2q35	***DES*** *125660*	Desmin	Li et al. (1999)	allelic to formerly LGMD1 related to *DES* and formerly LGM2R (group 1), MFM1 with or withour ARCV7 (group 5) and ARVC7 (group 10B)
Dilated cardiomyopathy, 1J	10.45	AD	CMD1J 605362	6q23.2	***EYA4*** *603550*	Eyes absent homolog	Schönberger et al. (2005)	
Dilated cardiomyopathy, 1K	10.46	AD	CMD1K 605582	6q12-q16	*?*	?	Sylvius et al. (2001)	
Dilated cardiomyopathy, 1L	10.47	AD	CMD1L 606685	5q33.2-q33.3	***SGCD*** *601411*	Sarcoglycan, delta	Tsubata et al. (2000)	allelic to LGMDR6 (group 1)
Dilated cardiomyopathy, 1M	10.48	AD	CMD1M 607482	11p15.1	***CSRP3*** *600824*	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Knoll et al. (2002)	allelic to CMH12 (group 10A)
Dilated cardiomyopathy, 1N	10.49	AD	CMD1N	17q12	***TCAP*** *604488*	Titin-cap (Telethonin)	Knoll et al. (2002)	allelic to LGMDR7 (group 1), CMD related to *TCAP* (group 2), CMH25 (group 10A)
Dilated cardiomyopathy, 1O	10.50	AD	CMD1O 605569	12p12.1	***ABCC9*** *601439*	ATP-binding cassette, subfamily C, member 9	Bienengraeber etal (2004)	allelic to ATFB12 (group 10B)
Dilated cardiomyopathy, 1P	10.51	AD	CMD1P 609909	6q22.31	***PLN*** *172405*	phospholamban	Schmitt et al. (2003) Haghighi et al. (2003, 2006)	allelic to CMH18 (group 10A)
Dilated cardiomyopathy, 1Q	10.52	AD	CMD1Q 609915	7q22.3-q31.1	*?*	?	Schonberger et al. (2005)	
Dilated cardiomyopathy, 1R	10.53	AD	CMD1R 613424	15q14	***ACTC1*** *102540*	actin, alpha, cardiac muscle	Olson et al. (1998) Mogensen et al. (1999)	allelic to CMH11, LVNC4 (group 10A)
Dilated cardiomyopathy, 1S	10.54	AD	CMD1S 613426	14q11.2	***MYH7*** *160760*	Myosin heavy chain 7 (beta), cardiac muscle	Kamisago et al. (2000)	allelic to CFTD, MSMA, MSMB, SPMM, eccentric core disease (group 3), MPD1 (group 4), CMH1 and LVNC5 (group 10A)
Dilated cardiomyopathy, 1T	10.55	AD	CMD1T	12q23.1	***TMPO*** *188380*	Thymopoietin (lamina-associated polypeptide 2)	Taylor et al. (2005)	
Dilated cardiomyopathy, 1U	10.56	AD	CMD1U 613694	14q24.2	***PSEN1*** *104311*	Presenilin 1	Li et al. (2006)	
Dilated cardiomyopathy, 1V	10.57	AD	CMD1V 613697	1q42.13	***PSEN2*** *600759*	Presenilin 2	Li et al. (2006)	

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Dilated cardiomyopathy, 1W	10.58	AD	CMD1W 611407	10q22.2	***VCL*** *193065*	Vinculin	Olson et al. (2002) Vasile et al. (2006)	allelic to CMH15 (group 10)
Dilated cardiomyopathy related, 1X	10.59	AR	CMD1X 611615	9q31.2	***FKTN*** *607440*	Fukutin	Murakami et al. (2006)	allelic to LGMDR13 (group 1), FCMD/MDDGA4 and WWS/MDDGB4 (group 2)
Dilated cardiomyopathy, 1Y	10.60	AD	CMD1Y 611878	15q22.2	***TPM1*** *191010*	Tropomyosin-1	Olson et al. (2010)	allelic to CMH3 and LVNC9 (group 10A)
Dilated cardiomyopathy, 1Z	10.61	AD	CMD1Z 611879	3p21.1	***TNNC1*** *191040*	Slow troponin C	Mogensen et al. (2004)	allelic to CMH13 (group 10A)
Dilated cardiomyopathy, 1AA, with or without LVNC	10.62	AD	CMD1AA 612158	1q42-q43	***ACTN2*** *102573*	Actinin alpha-2	Mohapatra et al. (2003)	allelic to MYOCOZ (group 3), MPD6 (group 4), CMH23 (group 10A)
Dilated cardiomyopathy, 1BB	10.63	AD/AR	CMD1BB 612877	18q12.1	***DSG2*** *125671*	Desmoglein 2	Posch et al. (2008)	allelic to ARVD10 (group 10B)
Dilated cardiomyopathy, 1CC	10.64	AD	CMD1CC 613122	1p31.1	***NEXN*** *613121*	Nexilin F-actin binding protein	Hassel et al. (2009)	allelic to CMH20 (group 10A)
Dilated cardiomyopathy, 1DD	10.65	AD	CMD1DD 613172	10q25.2	***RBM20*** *613171*	RNA-binding motif protein 20	Brauch et al. (2009)	
Dilated cardiomyopathy, 1EE	10.66	AD	CMD1EE 613252	14q11.2	***MYH6*** *160710*	Myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al. (2005)	allelic to CMH14 (group 10A), SSS3 (group 10B)
Dilated cardiomyopathy, 1FF	10.67	AD	CMD1FF 613286	19q13.42	***TNNI3*** *191044*	Cardic troponin I	Carballo et al. (2009)	allelic to CMH7, CDM2A, RCM1 (group 10A)
Dilated cardiomyopathy, 1GG	10.68	AR	CMD1GG 613642	5p15.33	***SDHA*** *600857*	Succinate dehydrogenase complex, subunit a, flavoprotein (**M**)	Levitas et al. (2010)	
Dilated cardiomyopathy, 1HH	10.69	AD	CMD1HH 613881	10q26.11	***BAG3*** *603883*	Bcl2-associated athanogene 3	Norton et al. (2011)	allelic to MFM6 (group 5) and CMT related to BAG3 (group 14)
Dilated cardiomyopathy, 1II	10.70	AD	CMD1II 615184	11q23.1	***CRYAB*** *123590*	Alpha-B crystallin	Inagaki et al. (2006)	allelic to MFM2 (group 5)
Dilated cardiomyopathy, 1JJ	10.71	AD	CMD1JJ 615235	6q21	***LAMA4*** *600133*	Laminin, alpha-4	Knöll et al. (2007)	
Dilated cardiomyopathy, 1KK	10.72	AD	CMD1KK 615248	10q21.3	***MYPN*** *608517*	Myopalladin	Duboscq-Bidot (2008)	allelic to NEM11 (group 3), CMH22 and RCM4 (group 10A)
Dilated cardiomyopathy, 1LL	10.73	AD	CMD1LL 615373	1p36.32	***PRDM16*** *605557*	PR domain-containing protein 16	Arndt et al. (2013)	allelic to LVNC8 (group 10A)
Dilated cardiomyopathy, 1MM	10.74	AD	CMD1MM 615396	11p11.2	***MYBPC3*** *600958*	Myosin-binding protein C, cardiac	Hershberger et al. (2010)	allelic to congenital myopathy and fatal cardiomyopathy (group 3), CMH4 and LVNC10 (group 10A)
Dilated cardiomyopathy, 1NN	10.75	AD	CMD1NN 615916	3p25.2	***RAF1*** *164760*	V-Raf-1 murine leukemia viral oncogene homolog 1	Dhandapany et al. (2014)	
Dilated cardiomyopathy related to *ILK*	10.76	AD		11p15.4	***ILK*** *602366*	Integrin-linked kinase	Knöll et al. (2007)	
Dilated cardiomyopathy related to *ANKRD1*	10.77	AD		10q23.31	***ANKRD1*** *609599*	Ankyrin repeat domain 1 containing protein1	Duboscq-Bidot et al. (2009) Moulik et al. (2009)	Allelic to Hypertrophic cardiomyopathy related to cardiac ankyrin repeat domain protein (group 10A)
Dilated cardiomyopathy, 2A	10.78	AR	CMD2A 611880	19q13.42	***TNNI3*** *191044*	Cardiac troponin I	Carballo et al. (2009)	allelic to CMH7, CMD1FF, RCM1 (group 10A)
Dilated cardiomyopathy 2B	10.79	AR	CMD2B 614672	7q21.2	***GATAD1*** *614518*	GATA zinc finger domain-containing protein 1	Theis et al. (2011)	
Dilated cardiomyopathy, 3A	10.80	XR	CMD3A	Xq28	***TAZ*** *300394*	Tafazzin	Gedeon et al. (1995)	allelic to BTHS (group 10A)
Dilated cardiomyopathy, 3B	10.81	XR	CMD3B 302045	Xp21.2-p21.1	***DMD*** *300377*	Dystrophin	Muntoni et al. (1993) Milasin et al. (1996)	allelic to DMD (group 1)
Dilated cardiomyopathy related to nesprin-1	10.82	AD		6q25.2	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope protein 1 (nesprin-1)	Puckelwartz et al. (2010)	allelic to EDMD4 (group 1) SCAR8 (group 13), AMCM (group 16)
Dilated cardiomyopathy related to MURC	10.83	AD		9q31.1	***CAVIN4*** *617714*	Caveolae-associated protein 4 (MURC)	Rodriguez et al. (2011)	
Dilated cardiomyopathy related to *DOLK*	10.84	AR	CDGM1 610768	9q34.11	***DOLK*** *610746*	Dolichol kinase	Kranz et al. (2007) Lefeber et al. (2011)	

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Restrictive cardiomyopathies								
Restrictive cardiomyopathy, 1	10.85	AD	RCM1 115210	19q13.42	***TNNI3*** *191044*	Cardiac troponin I	Mogensen et al. (2003)	allelic to CMH7, CMD1FF, CMD2A (group 10A)
Restrictive cardiomyopathy, 2	10.86		RCM2 609578	10q23.3	*?*	?	Zhang et al. (2005)	
Restrictive cardiomyopathy, 3	10.87	AD	RCM3 612422	1q32.1	***TNNT2*** *191045*	Cardiac Troponin T	Peddy et al. (2006)	allelic to CMH2, CMD1D and LVNC6 (group 10a)
Restrictive cardiomyopathy, 4	10.88	AD	RCM4 615248	10q21.3	***MYPN*** *608517*	Myopalladin	Purevjav et al. (2012)	allelic to NEM11 (group 3), CMH22 and CMD1KK (group 10A)
Restrictive cardiomyopathy, 5	10.89	AD	RCM5 617047	7q32.1	***FLNC*** *102565*	Filamin C	Brodehl et al. (2016)	allelic to MFM5 (group 5), MPD4 (group 4) and CMH26 (group 10A)
Other non arrhythmogenic hereditary cardiomyopathies								
Pompe disease, Glycogenosis, generalized, cardiac form (*also listed in group 9*)	10.90	AR	GSD2 232300	17q25.3	***GAA*** *606800*	Glucosidase, alpha; acid	Hers (1963) Martiniuk et al. (1990) Wokke et al. (1995)	Allelic to Pompe disease (formerly LGMD2V) (group 1)
Cardioskeletal myopathy with neutropenia and abnormal mitochondria (Barth syndrome)	10.91	XR	BTHS 302060	Xq28	***TAZ*** *300394*	Tafazzin	Bolhuis et al. (1991) Bione et al. (1996)	allelic to CMD3A (group 10A)
Left ventricular noncompaction, 1	10.92	AD	LVNC1 604169	18q12.1	***DTNA*** *601239*	Dystrobrevin, alpha	Ichida et al. (2001)	
Left ventricular noncompaction, 2	10.93		LVNC2 609470	11q15	***?***	?	Sasse-Klaassen et al. (2004)	
Left ventricular noncompaction, 3	10.94	AD	LVNC3 601493	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Vatta et al. (2003)	alllelic to Late onset distal myopathy (group 4), MFM4 (group 5), CMH24 and CMD1C (group 10A)
Left ventricular noncompaction, 4	10.95	AD	LVNC4 613424	15q14	***ACTC1*** *102540*	Alpha-cardiac actin	Monserrat et al. (2007)	allelic to CMH11, CMD1R (group 10A)
Left ventricular noncompaction, 5	10.96	AD	LVNC5 613426	14q11.2	***MYH7*** *160760*	Myosin heavy chain 7 (beta), cardiac muscle	Klaassen et al. (2008)	allelic to CFTD, MSMA, MSMB, SPMM, eccentric core disease (group 3), MPD1 (group 4), CMH1 and CMD1S (group 10A)
Left ventricular noncompaction, 6	10.97	AD	LVNC6 601494	1q32.1	***TNNT2*** *191045*	Cardiac Troponin T	Luedde et al. (2010)	allelic to CMH2, CMD1D and RCM3 (group 10A)
Left ventricular noncompaction, 7	10.98	AD	LVNC7 615092	18q11.2	***MIB1*** *608677*	Mindbomb, drosophila, homolog of	Luxan et al. (2013)	
Left ventricular noncompaction, 8	10.99	AD	LVNC8 615373	1p36.32	***PRDM16*** *605557*	PR domain-containing protein 16	Arndt et al. (2013)	allelic to CMD1LL (group 10A)
Left ventricular noncompaction, 9	10.100	AD	LVNC9 611878	15q22.2	***TPM1*** *191010*	Tropomyosin-1	Probst et al. (2011)	allelic to CMH3, CMD1Y (group 10A)
Left ventricular noncompaction, 10	10.101	AD	LVNC10 615396	11p11.2	***MYBPC3*** *600958*	Cardiac myosin binding protein-C	Probst et al. (2011)	allelic to congenital skeletal myopathy and fatal cardiomyopathy (group 3), CMH4 and CMD1MM (group 10A)
Cardiovalvular dysplasia, X-linked (Myxomatous valvular dystrophy)	10.102	XR	CVD1 314400	Xq28	***FLNA*** *300017*	Filamin A, alpha (actin binding protein 280)	Kyndt et al. (1998) Kyndt et al. (2007)	
GROUP 11. CONGENITAL MYASTHENIC SYNDROMES								
Myasthenic syndrome, congenital, 1A, Slow-channel	11.1	AD	CMS1A 601462	2q31.1	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1	Sine et al. (1995) Engel et al. (1996) Croxen et al. (1997)	allelic to CMS1B (group 11)
Myasthenic syndrome, congenital, 2A, Slow-channel	11.2	AD	CMS2A 616313	17p13.1	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1	Engel et al. (1996b) Gomez et al. (1996)	allelic to CMS2B, CMS2C (group 11)
Myasthenic syndrome, congenital, 3A, Slow-channel	11.3	AD	CMS3A 616321	2q37.1	***CHRND*** *100720*	Cholinergic receptor, Cicotinic, delta	Gomez et al. (2002)	allelic to CMS3B, CMS3C (group 11)
Myasthenic syndrome, congenital, 4A, Slow-channel	11.4	AD, AR	CMS4A 605809	17p13.2	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Ohno et al. (1995) Gomez et al. (1995) Engel et al. (1996) Croxen et al. (2002)	allelic to CMS4B, CMS4C (group 11)

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Myasthenic syndrome, congenital, 1B, Fast-channel	11.5	AR	CMS1B 608930	2q31.1	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1	Wang et al. (1999) Shen et al. (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 2B, Fast-channel	11.6	AR	CMS2B	17p13.1	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1	Shen et al. (2020)	allelic to CMS2A, CMS2C (group 11)
Myasthenic syndrome, congenital, 3B, Fast-channel	11.7	AR	CMS3B 616322	2q37.1	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Brownlow et al. (2001)	allelic to CMS3A, CMS3C (group11)
Myasthenic syndrome, congenital, 4B, Fast-channel	11.8	AR	CMS4B 616324	17p13.2	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Ohno et al. (1996)	allelic to CMS4A, CMS4C (group 11)
Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	11.9	AR	CMS2C 616314	17p13.1	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1	Quiram et al. (1999)	allelic to CMS2A, CMS2B (group 11)
Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	11.10	AR	CMS3C 616323	2q37.1	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Shen et al. (2002)	allelic to CMS3A, CMS3B (group11)
Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	11.11	AR	CMS4C 608931	17p13.2	***CHRNE*** *100725*	Ccholinergic receptor, nicotinic, epsilon	Engel et al. (1996) Ohno et al. (1997)	allelic to CMS4A, CMS4B (group 11)
Myasthenic syndrome, congenital, 5	11.12	AR	CMS5 603034	3p25.1	***COLQ*** *603033*	Collagenic tail of endplate acetylcholinesterase	Donger et al. (1998) Ohno et al. (1998–2000)	
Myasthenic syndrome, congenital, 6, presynaptic	11.13	AR	CMS6 254210	10q11.23	***CHAT*** *118490*	Choline acetyltransferase	Ohno et al. (2001) Maselli et al. (2003)	
Myasthenic syndrome, congenital, 7, presynaptic	11.14	AD	CMS7 616040	1q32.1	***SYT2*** *600104*	Synaptotagmin 2	Herrmann et al. (2014)	Allelic to CMS7B (group 11) and to Distal motor neuropathy related to SYT2 (group 12)
Myasthenic syndrome, congenital, 7B, presynaptic	11.15	AR	CMS7B 619461	1q32.1	***SYT2*** *600104*	Synaptotagmin 2	Masseli et al. (2020)	Allelic to CMS7 (group 11) and to Distal motor neuropathy related to SYT2 (group 12)
Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	11.16	AR	CMS8 615120	1p36.33	***AGRN*** *103320*	Agrin	Huzé et al. (2009)	Allelic to FADS related to *AGRN* (group 16)
Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	11.17	AR	CMS9 616325	9q31.3	***MUSK*** *601296*	Muscle-specific receptor tyrosine kinase	Chevessier et al. (2004)	allelic to FADS (group 16)
Myasthenic syndrome, congenital, 10	11.18	AR	CMS10 254300	4p16.3	***DOK7*** *610285*	Downstream of tyrosin kinase 7	Beeson et al. (2006) Selcen et al. (2008)	allelic to FADS (group 16)
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	11.19	AR	CMS11 616326	11p11.2	***RAPSN*** *601592*	Receptor-associated protein of the synapse, 43 kD (Rapsyn)	Ohno et al. (2002) Ohno et al. (2003) Dunne et al. (2003)	allelic to FADS (group 16)
Myasthenia, congenital, 12, with tubular aggregates	11.20	AR	CMS12 610542	2p13.3	***GFPT1*** *138292*	Glutamine:fructose-6-phosphate amido transtransferase 1	Senderek et al. (2011)	
Myasthenic syndrome, congenital, 13, with tubular aggregates	11.21	AR	CMS13 614750	11q23.3	***DPAGT1*** *191350*	Dolichyl-phosphate N-acetylglucosamine phosphotransferase 1	Belaya et al. (2012)	
Myasthenic syndrome, congenital, 14, with tubular aggregates	11.22	AR	CMS14 616228	9q22.33	***ALG2*** *607905*	S. Cerevisae homolog of ALG2 (alpha-1,3/1,6-mannosyl transferase)	Cossins et al. (2013)	
Myasthenic syndrome, congenital, 15, without tubular aggregates	11.23	AR	CMS15 607227	1p21.3	***ALG14*** *612866*	S. Cerevisae homolog of ALG14 (UDP-N-acetylglucosami-nyltransferase subunit)	Cossins et al. (2013)	
Myasthenic syndrome, congenital, 16	11.24	AR	CMS16 614198	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subnit	Tsujino et al. (2003)	allelic to Congenital Myopathy related to *SCN4A* (group 3), HOKPP2, HYPP, PMC and K-aggravated myotonia (group 7), Severe foetal hypokinesia related to *SCN4A* (group 16)
Myasthenic syndrome, congenital, 17	11.25	AR	CMS17 616304	11p11.2	***LRP4*** *604270*	LDL receptor-related protein 4	Ohkawara et al. (2014)	
Myasthenic syndrome, congenital, 18	11.26	AD	CMS18 616330	20p12.2	***SNAP25*** *600322*	Synaptosomal associated protein 25	Shen et al. (2014)	

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Myasthenic syndrome, congenital, 19	11.27	AR	CMS19 616720	10q22.1	***COL13A1*** *120350*	Collagen type XIII alpha 1 chain	Logan et al. (2015)	
Myasthenic syndrome, congenital, 20, presynaptic	11.28	AR	CMS20 617143	2q12.3	***SLC5A7*** *608761*	Solute carrier family 5 (choline transporter) member 7	Bauche et al. (2016)	allelic to HMN7A (group 12)
Myasthenic syndrome, congenital, 21, presynaptic	11.29	AR	CMS21 617239	10q11.23	***SLC18A3*** *600336*	Solute carrier family 18 (vesicular acetylcholine), member 3	O'Grady et al. (2016)	
Myasthenic syndrome, congenital, 22	11.30	AR	CMS22 616224	2p21	***PREPL*** *609557*	Prolyl endopeptidase-like	Regal et al. (2014)	
Presynaptic congenital myasthenic syndrome 23	11.31	AR	CMS23 618197	22q11.21	***SLC25A1*** *190315*	Solute carrier family 25 (mitochondrial carrier, citrate transporter), member 1	Chaouch et al. 2014)	
Presynaptic congenital myasthenic syndrome 24	11.32	AR	CMS24 618198	15q23	***MYO9A*** *604875*	Myosin IXA	O'Connor et al. (2016)	
Presynaptic congenital myasthenic syndrome 25	11.33	AR	CMS25 618323	12p13.31	***VAMP1*** *185880*	Vesicle-associated membrane protein 1	Shen et al. (2017) Salpieto et al. (2017)	allelic to SPAX1 (group 15)
Congenital myasthenic syndrome with nephrotic syndrome	11.34	AR	NPHS5 614199	3p21.31	***LAMB2*** *150325*	Laminin-beta 2	Maselli et al. (2009)	
Escobar syndrome (multiple pterygium syndrome)	11.35	AR	EVMPS 265000	2q37.1	***CHRNG*** *100730*	Cholinergic receptor, nicotinic, gamma	Hoffman et al. (2006) Morgan et al. (2006)	
Myasthenic syndrome, with plectin defect	11.36	AR		8q24.3	***PLEC*** *601282*	Plectin	Banwell et al. (1999) Forrest et al. (2010) Selcen et al. (2011)	Allelic to LGMDR17 (group 1), LGMD with ophthalmoplegia (group 1) and EBSMD (group 5)
Congenital myasthenic syndrome related to *GMPPB*	11.37	AR		3q21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Belaya et al. (2015)	allelic to LGMDR19 (group 1); MEB/MDDGA14 and MDDGB14 (group 2)
Presynaptic congenital myasthenic syndrome	11.38	AR		20q13.33	***LAMA5*** *601033*	Laminin, alpha-5	Maselli et al. (2017)	
Presynaptic congenital myasthenic syndrome related to MUNC13–1	11.39	AR		19p13.11	***UNC13A*** *609894*	C. Elegans, homolog of UNC13A (MUNC13)	Engel et al. (2016)	
Congenital myasthenic syndrome related to RPH3A, presynaptic	11.40	AR		12q24.13	***RPH3A*** *612159*	Rabphilin 3A	Maselli et al. (2018)	
Congenital myasthenic syndrome	11.41	AR		1q25.2	***TOR1AIP1*** *(=LAP1B) 614512*	Torsin A interacting protein 1 (=Lamin Associated Peptide 1B)	Cossins et al. (2020) Malfatti et al. (2021)	Allelic to MRRSDC (group 1)
Congenital myasthenic syndrome	11.42	AD		14q11.2	***CHD8*** *610528*	Chromodomain Helicase DNA-Binding Protein 8	Lee et al. (2020)	
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	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Gilliam et al. (1990) Melki et al. (1990, 1994) Lefebvre et al. (1995) Bussaglia et al. (1995) Rodrigues et al. (1995) Roy et al. (1995) Hahnen et al. (1997)	allelic to SMA2, SMA3, SMA4 (group 12)
Spinal muscular atrophy, type II (intermediate)	12.2	AR	SMA2 253550	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Matthijs et al. (1996) Samilchuk (1996)	allelic to SMA1, SMA3, SMA4 (group 12)
Spinal muscular atrophy, type III (Kugelberg-Welander)	12.3	AR	SMA3 253400	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Brzustowicz et al. (1990) Melki et al. (1990b) Lefebvre et al. (1995)	allelic to SMA2, SMA1, SMA4 (group 12)
Spinal muscular atrophy, type IV, adult form	12.4	AR	SMA4 271150	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Brahe et al. (1995) Clermont et al. (1995)	allelic to SMA2, SMA3, SMA1 (group 12)

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Distal spinal muscular atrophy, recessive								
Spinal muscular atrophy, distal, autosomal recessive 1 (with respiratory distress)	12.5	AR	DSMA1 604320	11q13.3	***IGHMBP2*** *600502*	Immunoglobulin Mu-binding protein 2	Grohmann et al. (1999, 2001)	allelic to CMT2S (group 14)
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	1p36.31	***PLEKHG5*** *611101*	Pleckstrin homology domain and RhoGEF domai-containing protein G5	Maystadt et al. (2006, 2007)	allelic to CMTRIC (group 14)
Spinal muscular atrophy, distal, autosomal recessive, 5	12.9	AR	DSMA5 614881	2q35	***DNAJB2*** *604139*	DnaJ/Hsp40 homolog, subfamily B, member 2	Blumen et al. (2012)	Allelic to CMT related to DNAJB2 (group 14)
Spinal muscular atrophy, distal, autosomal recessive	12.10	AR	DSMA	14q32.2	***VRK1*** *602168*	Vaccinia related kinase 1	Stoll et al. (2016) Li et al. (2019)	Allelic to PCH1 (group 12), Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis (group 14)
Spinal muscular atrophy with congenital bone fractures 1	12.11	AR	SMABF1 616866	15q22.31	***TRIP4*** *604501*	Thyroid hormone receptor interactor 4	Knierim et al. (2016)	allelic to MDCDC (group 2)
Spinal muscular atrophy with congenital bone fractures 2	12.12	AR	SMABF2 616867	10q22.1	***ASCC1*** *614215*	Activating signal cointegrator 1 complex subunit 1	Knierim et al. (2016)	allelic to Arthrogryposis related to *ASCC1*(group 16)
Spinal muscular atrophy with progressive myoclonic epilepsy	12.13	AR	SMAPME 159950	8p22	***ASAH1*** *613468*	N-acylsphingosine amidohydrolase 1	Zhou et al. (2012)	allelic to FRBRL (#228000)
Spinal muscular atrophy and cerebellar hypoplasia	12.14	AR	PCH1C 616081	13q13.3	***EXOSC8*** *606019*	Exosome component 8	Boczonadi et al. (2014)	
Distal hereditary motor neuronopathy	12.15	AR		5q23.3	***HINT1*** *601314*	Histidine triad nucleotide-binding protein	Zhao et al. (2014)	Allelic to NMAN (group 14)
Distal spinal muscular atrophy, dominant								
Neuronopathy, distal hereditary motor, type I	12.16	AD	HMN1 182960	7q34-q36	***?***	?	Gopinath et al. (2007)	
Neuronopathy, distal hereditary motor, type IIA	12.17	AD	HMN2A 158590	12q24.23	***HSPB8*** *608014*	Heat-shock 22-kD protein 8	Timmerman et al. (1992) Irobi et al. (2004)	allelic to Rimmed vacuole myopathy and distal myopathy and motor neuropathy (group 4) and CMT2L (group 14)
Neuronopathy, distal hereditary motor, type IIB	12.18	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	12.19	AD	HMN2C 613376	5q11.2	***HSPB3*** *604624*	Heat shock 27-kD protein 3	Kolb et al. (2010)	
Neuronopathy, distal hereditary motor, type IID	12.20	AD	HMN2D 615575	5q32	***FBXO38*** *608533*	F-box only protein 38	Sumner et al. (2013)	
Distal spinal muscular atrophy, distal with upper limb predominance (type V)	12.21	AD	HMN5A 600794	7p14.3	***GARS*** *600287*	Glycyl-tRNA synthetase	Christodoulou et al. (1995) Antonellis et al. (2003)	allelic to CMT2D (group 14)
Distal spinal muscular atrophy type VA	12.22	AD	HMN5A 600794	11q12.3	***BSCL2*** *606158*	Seipin	Windpassinger et al. (2004)	allelic to SPG17 (group 15)
Distal spinal muscular atrophy, type VB	12.23	AD	HMNS5B 614751	2p11.2	***REEP1*** *609139*	Receptor expression-enhancing protein 1	Beetz et al. (2012)	allelic to SPG31 (group 15)
Dominant distal hereditary motor neuropathy	12.24	AD	dHMN	16q22.1	***AARS1*** *601065*	Alanyl-tRNA synthetase 1	Zhao et al. (2012)	allelic to CMT2N (group 14)
Neuronopathy, distal hereditary motor, type IX	12.25	AD	HMN9 61772	14q32.2	***WARS1*** *191050*	Tryptophanyl-tRNA synthetase 1	Tsai et al. (2017)	
Spinal muscular atrophy, distal, with vocal cord paralysis (Harper-Young)	12.26	AD	HMN7A 158580	2q12.3	***SLC5A7*** *608761*	Solute carrier family 5 (choline cotransporter), member 7	McEntagart et al. (2001) Barwick et al. (2012)	allelic to CMS20 (group 11)
Distal hereditary motor neuronopathy type VIIB	12.27	AD	HMN7B 607641	2p13.1	***DCTN1*** *601143*	Dynactin 1	Puls et al. (2003)	allelic to ALS related to DCTN1 (group 12)
Hereditary motor and sensory neuropathy V	12.28	AD	HMSN5 600361	4q34.3-q35.2	*?*	?	Muglia et al. (2008)	

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Spinal muscular atrophy, distal, congenital non progressive	12.29	AD	HMN8 600175	12q24.11	***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 12), HSMN2C (group 14)
Scapuloperoneal spinal muscular atrophy	12.30	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 HMN8 (group 12), HSMN2C (group 14)
Spinal motor neuropathy	12.31			11q23.2	***RBM7*** *612413*	RNA-binding motif protein 7	Giunta et al. (2016)	
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 1	12.32	AD	SMALED1 158600	14q32.31	***DYNC1H1*** *600112*	Dynein, cytoplasmic 1, heavy chain 1	Harms et al. (2010, 2012)	allelic to CMT2O (group 14)
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 2A	12.33	AD	SMALED2A 615290	9q22.31	***BICD2*** *609797*	Bicaudal D homolog 2 (Drosophila)	Neveling et al. (2013) Oates et al. (2013) Peeters et al. (2013)	Allelic to SMALED2B (group 12) and Arthrogryposis and BICD2-related neuromuscular disease (group 16)
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 2 B	12.34	AD	SMALED2B 618291	9q22.31	***BICD2*** *609797*	Bicaudal D homolog 2 (Drosophila)	Ravenscroft et al. (2016)	Allelic to SMALED2A (group 12) and Arthrogryposis and BICD2-related neuromuscular disease (group 16)
Spinal muscular atrophy, late-onset, Finkel type	12.35	AD	SMAFK 182980	20q13.32	***VAPB*** *605704*	Vesicle-associated membrane protein-associated protein B	Nishimura et al. (2004)	allelic to ALS8 (group 12)
Spinal muscular atrophy, Jokela type	12.36	AD	SMAJ 615048	22q11.23	***CHCHD10*** *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), IMMD (group 16)
Distal motor neuropathy related to SYT2	12.37	AD		1q32.1	***SYT2*** *600104*	Synaptotagmin 2	Montes-Chinea et al. (2018)	Allelic to CMS7, CMS7B (group 11)
Distal motor neuropathy	12.38	AD		9q34.11	***SPTAN1*** *182810*	Spectrin, alpha, nonerythrocytic 1	Beijer et al. (2019)	
Distal hereditary motor neuropathies	12.39	AD		10q24.32	***GBF1*** *603698*	Golgi-specific brefeldin-A resistance factor 1	Mendoza-Ferreira et al. (2020)	Allelic to CMT2GG (group 14)
Distal spinal muscular atrophy, X-linked								
Spinal and bulbar muscular atrophy, X-linked, 1 (Kennedy disease)	12.40	XR	SBMX1 313200	Xq12	***AR*** *313700*	Androgen receptor	Fishbeck et al. (1986) La Spada et al. (1991)	
Spinal muscular atrophy, distal, X-linked 2	12.41	XR	SMAX2 301830	Xp11.3	***UBA1*** *314370*	Ubiquitin-like modifyer-activating enzyme 1	Ramser et al. (2013) Diamini et al. (2013)	
Spinal muscular atrophy, distal, X-linked, 3	12.42	XR	SMAX3 300489	Xq21.1	***ATP7A*** *300011*	ATPase, Cu^{++}transporting, alpha polypeptide	Takata et al. (2004) Kennerson et al. (2010)	
Amyotrophic lateral sclerosis (ALS)								
Amyotrophic lateral sclerosis 1 (dominant)	12.43	AD	ALS1 105400	21q22.11	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Siddique et al. (1991, 1996) Rosen et al. (1993)	
Amyotrophic lateral sclerosis 1 (recessive)	12.44	AR	ALS1 105400	21q22.11	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Andersen et al. (1995)	
Amyotrophic lateral sclerosis 2, juvenile	12.45	AR	ALS2 205100	2q33.1	***ALS2*** *606352*	Alsin Rho guanine nucleotide exchange factor 2	Hentati et al. (1994) Yang et al. (2001) Hadano et al. (2001)	allelic to IAHSP (group 15)
Amyotrophic lateral sclerosis 3	12.46	AD	ALS3 606640	18q21	*?*	?	Hand et al. (2002)	
Amyotrophic lateral sclerosis 4, juvenile	12.47	AD	ALS4 602433	9q34.13	***SETX*** *608465*	Senataxin	Chance et al. (1998) Chen et al. (2004) Moreira et al. (2004)	allelic to SCAR1 (group 13)
Amyotrophic lateral sclerosis 5, juvenile	12.48	AR	ALS5 602099	15q21.1	***SPG11*** *610844*	Spatacsin	Hentati et al. (1998) Orlacchio et al. (2010)	allelic to CMT2X (group 14)
Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	12.49	AD	ALS6 608030	16p11.2	***FUS*** *137070*	Fused in sarcoma	Sapp et al. (2003) Abalkhail et al. (2003) Kwiatkowski et al. (2009) Vance et al. (2009)	

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Amyotrophic lateral sclerosis 7	12.50	AD	ALS7 608031	20p13	*?*	?	Sapp et al. (2003)	
Amyotrophic lateral sclerosis 8	12.51	AD	ALS8 608627	20q13.32	***VAPB*** *605704*	Vesicle-associated membrane protein-associated protein B and C	Nishimura et al. (2004)	allelic to SMAFK (group 12)
Amyotrophic lateral sclerosis 9	12.52	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	12.53	AD	ALS10 612069	1p36.22	***TARDBP*** *605078*	TAR DNA-binding protein	Sreedharan et al. (2008)	
Amyotrophic lateral sclerosis 11	12.54	AD	ALS11 612577	6q21	***FIG4*** *609390*	FIG4, S. Cerevisiae, homolog of	Chow et al. (2009)	allelic to CMT4J (group 14)
Amyotrophic lateral sclerosis 12	12.55	AD, AR	ALS12 613435	10p13	***OPTN*** *602432*	Optineurin	Maruyama et al. (2010)	
Amyotrophic lateral sclerosis 13	12.56	AD	ALS13 183090	12q24.12	***ATXN2*** *601517*	Ataxin 2	Elden et al. (2010) Daoud et al. (2011) Van Damme et al. (2011)	allelic to SCA2 (group 13)
Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia	12.57	AD	ALS14 613954	9p13.3	***VCP*** *601023*	Valosin-containing protein	Johnson et al. (2011)	allelic to Scapuloperoreal myopathy (group 1), IBMPFD1 (groups 4 and 5), CMT2Y (group 14)
Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	12.58	XD	ALS15 300857	Xp11.21	***UBQLN2*** *300264*	Ubiquilin 2	Deng et al. (2011)	
Amyotrophic lateral sclerosis 16, juvenile	12.59	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	12.60	AD	ALS17 614696	3p11.2	***CHMP2B*** *609512*	Charged multivesicular body protein 2B	Parkinson et al. (2006) Cox et al. (20010)	
Amyotrophic lateral sclerosis 18	12.61	AD	ALS18 614808	17p13.2	***PFN1*** *176610*	Profilin 1	Wu et al. (2012)	
Amyotrophic lateral sclerosis 19	12.62	AD	ALS19 615515	2q34	***ERBB4*** *600543*	V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 4	Takahashi et al. (2013)	
Amyotrophic lateral sclerosis 20	12.63	AD	ALS20 615426	12q13.13	***HNRNPA1*** *164017*	Heterogeneous nuclear ribonucleoprotein A1	Kim et al. (2013)	allelic to IBMPFD3 (group 3)
Amyotrophic lateral sclerosis 21	12.64	AD	ALS21 606070	5q31.2	***MATR3*** *164015*	Matrin 3	Johnson et al. (2014)	allelic to VCPDM (group 4)
Amyotrophic lateral sclerosis 22, with or without frontotemporal dementia	12.65	AD	ALS22 616208	2q35	***TUBA4A*** *191110*	Tubulin, alpha-4A	Smith et al. (2014)	
Amyotrophic lateral sclerosis 23	12.66	AD	ALS23 617839	10q22.3	***ANXA11*** *602572*	Annexin A11	Smith et al. (2017)	allelic to Multisystem proteinopathy (group 12)
Amyotrophic lateral sclerosis 24, suseptibility to	12.67		ALS24 617892	4q33	***NEK1*** *604588*	Never in mitosis gene A-related kinase 1	Brenner et al. (2016) Kenna et al. (2016)	
Amyotrophic lateral sclerosis 25, susceptibility to	12.68	AD	ALS25 617921	12q13.3	***KIF5A*** *602821*	Kinesin family member 5A	Nicolas et al. (2018)	Allelic to CMT2 related to KIF5A (group 14) and SPG10 (group 15)
Amyotrophic lateral sclerosis related to NEFH, susceptibility to	12.69	AD, AR	ALS1 105400	22q12.2	***NEFH*** *162230*	Neurofilament, heavy polypeptide	Al-Chalabie et al. (1999)	allelic to CMT2CC (group 14)
Amyotrophic lateral sclerosis related to peripherin, susceptibility to	12.70	AD	ALS1 105400	12q13.12	***PRPH*** *170710*	Peripherin	Gros-Louis et al. (2004) Leung et al. (2004)	
Amyotrophic lateral sclerosis related to dynactin 1, susceptibility to	12.71	AD	ALS1 105400	2p13.1	***DCTN1*** *601143*	Dynactin 1	Munch et al. (2005)	allelic to HMN7B (group 12)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.72	AD	FTDALS1 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	12.73	AD	FTDALS2 615911	22q11.23	***CHCHD10*** *615903*	Coiled-coil-helix-coiled-coil-helix domain containing 10 (**M**)	Bannwarth et al. (2014)	allelic to SMAJ (group 12), IMMD (group 16)

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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)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.74	AD	FTDALS3 66437	5q35.3	***SQSTM1*** *601530*	Sequestosome 1	Fecto et al. (2011)	allelic to Welander-like syndrome and DMRV (group 4)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.75	AD	FTDALS4 616439	12q14.2	***TBK1*** *604834*	Tank-binding kinase 1	Cirulli et al. (2015) Freischmidt et al. (2015)	
Amyotrophic lateral sclerosis	12.76	AD		14q11.2	***LRP10*** *609921*	Low Density Lipoprotein Receptor-Related Protein 10	Ni et al. (2021)	
Amyotrophic lateral sclerosis	12.77	AD		9q22.31	***SPTLC1*** *605712*	Serine palmitoyl transferase long-chain base subunit 1	Mohassel et al. (2021)	Allelic to HSAN1A (group 14)
Others								
Lethal Congenital Contracture Syndrome 1	12.78	AR	LCCS1 253310	9q34.11	***GLE1*** *603371*	GLE1, RNA export mediator	Makela-Bengs et al. (1998) Nousiainen et al. (2008)	
Lethal Congenital Contracture Syndrome 2	12.79	AR	LCCS2 607598	12q13.2	***ERBB3*** *190151*	V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 3	Narkis et al. (2007)	
Lethal Congenital Contracture Syndrome 3	12.80	AR	LCCS3 611359	19p13.3	***PIP5K1C*** *606102*	Phosphatidylinositol-4-phosphate 5-kinase, type I, gamma	Narkis et al. (2007)	
Lethal Congenital Contracture Syndrome 4	12.81	AR	LCCS4 614915	12q23.2	***MYBPC1*** *160794*	Myosin-binding protein C, slow type	Markus et al. (2012)	Allelic to DA1B (group 16)
Lethal Congenital Contracture Syndrome 5	12.82	AR	LCCS5 615368	19p13.2	***DNM2*** *602378*	Dynamin 2	Koutsopoulos et al. (2013)	Allelic to CMD related to *DNM2* (group 2), CNM1 (group 3), Distal myopathy related to *DNM2* (group 4), and CMTDIB (group 14)
Lethal Congenital Contracture Syndrome 6	12.83	AR	LCCS6 616248	14q32.33	***ZBTB42*** *613915*	Zinc finger and BTB domain-containing protein 42	Patel et al. (2014)	
Lethal Congenital Contracture Syndrome 7	12.84	AR	LCCS7 616286	17q21.2	***CNTNAP1*** *602346*	Contactin-associated protein 1	Laquerriere et al. (2014) Lakhani et al. (2017)	Allelic to CMT related to *CNTNAP1* (group 14)
Lethal Congenital Contracture Syndrome 8	12.85	AR	LCCS8 616287	12q13.12	***ADCY6*** *600294*	Adenylate cyclase 6	Laquerriere et al. (2014) Agolini et al. (2020)	
Lethal Congenital Contracture Syndrome 9	12.86	AR	LCCS9 616503	6q24.2	***ADGRG6*** *612243*	Adhesion G protein-coupled recptor G6	Ravenscroft et al. (2015)	
Lethal Congenital Contracture Syndrome 10	12.87	AR	LCCS10 617022	14q24.3	***NEK9*** *609798*	Never in mitosis gene A-related kinase 9	Casey et al. (2016)	
Lethal Congenital Contracture Syndrome 11	12.88	AR	LCCS11 617194	15q21.2	***GLDN*** *608603*	Gliomedin	Maluenda et al. (2016)	
Lethal Congenital Contracture Syndrome related to *TTN*	12.89	AR		2q31.2	***TTN*** *188840*	Titin	Chervinski et al. (2018)	Allelic to LGMDR10 (group 1), CNM related to *TTN* (group 3), MmD related to *TTN* (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10), CMD1G (group 10)
Spinal muscular atrophy with pontocerebellar hypoplasia, type 1b	12.90	AR	PCH1B 614678	9p13.2	***EXOSC3*** *606489*	Exosome component 3	Rudnik-Schöneborn et al. (2013)	
Spinal muscular atrophy with or without pontocerebellar hypoplasia	12.91	AR	PCH1 607596	14q32.2	***VRK1*** *602168*	Vaccinia related kinase 1	Renbaum et al. (2009) Stoll et al. (2016)	Allelic to DSMA (group 12), Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis (group 14)
Pontocerebellar hypoplasia with spinal muscular atrophy	12.92	AR	HSMN6B 616505	5q22.1	***SLC25A46*** *610826*	Solute Carrier Family 25, Member 46	van Dijk et al. (2017)	
Brown-Vialetto-van Laere syndrome 1	12.93	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	12.94	AR	BVVLS2 614707	8q24.3	***SLC52A2*** *607882*	Solute carrier family 52 (riboflavin transporter) member 2	Green et al. (2010) Bosch et al. (2011) Johnson et al. (2012)	

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Late onset spinal muscular atrophy related to *HEXB*	12.95	AR		5q13.3	***HEXB*** *606873*	Hexosaminidase B	Rattay et al. (2013)	allelic to Sandhoff disease, adult type (OMIM #268800)
Spinal muscular atrophy, related to *PRUNE1*	12.96	AR		1q21.3	***PRUNE1*** *617413*	Prune exopolyphosphatase 1	Iacomino et al. (2017)	
Spinal muscular atrophy with pontocerebellar hypoplasia related to *KIF26B*	12.97	AD		1q44	***KIF26B*** *614026*	Kinesin family member 26B	Wojcik et al. (2018)	
Peripheral neuropathy, myopathy, hoarseness and hearing loss	12.98	AD	PNMHH 614369	19q13.33	***MYH14*** *608568*	Myosin, heavy chain 14, non muscle	Choi et al. (2011)	
Lower motor neuron disease with respiratory failure related to *MAPT*	12.99	AD		17q21.31	***MAPT*** *157140*	Microtubule associated protein Tau	Di Fonso et al. (2014)	allelic to Frontotemporal dementia (OMIM #600274)
Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	12.100	AR	PNRIID 618124	21q22.3	***MCM3AP*** *603294*	Minichromosome maintenance 3-associated protein	Schuurs-Hoeijmakers et al. (2013) Ylikallio et al. (2017)	allelic to CMT2 (group 14)
Neuropathy, hereditary motor, with myopathic features	12.101	AR	HMNMYO 619216	1p36.33	***VWA1*** *611901*	Von Willebrand factor A domain-containing protein 1	Deschauer et al. (2021) Pagnamenta et al. (2021)	
Multisystem proteinopathy	12.102	AD		10q22.3	***ANXA11*** *602572*	Annexin A11	Leoni et al. (2021)	allelic to ALS23 (group 12)
Sensory neuropathy	12.103	AR		1q44	***COX20*** *614698*	Cytochrome c Oxidase Assembly Factor COX20	Dong et al. (2021)	
Progressive neuropathy	12.104	AR		9q21.2	***PSAT1*** *610936*	Phosphoserine Aminotransferase 1	Debs et al. (2021)	
Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures	12.105	AR	CONDSIAS 618170	1p34.3	***ADPRHL2*** *610624*	ADP-Ribosylhydrolase-Like 2	Danhauser et al. (2018) Ghosh et al. (2018)	
GROUP 14. HEREDITARY MOTOR SENSORY NEUROPATHIES (HMSN)								
A. Charcot-Marie-Tooth neuropathy, type 1 (demyelinating)								
Autosomal dominant (AD-CMT1)								
Charcot-Marie-Tooth disease, type 1A	14.1	AD	CMT1A 118220	17p12	***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. (1993)	allelic to CMT1E, HNPP and DSS (group 14)
Charcot-Marie-Tooth disease, type 1B	14.2	AD	CMT1B 118200	1q23.3	***MPZ*** *159440*	Myelin protein zero	Bird et al. (1982) Guiloff et al. (1982) Hayasaka et al. (1993) Kulkens et al. (1993)	allelic to CMTDID, CMT2I, CMT2J, DSS and CHN (group 14)
Charcot-Marie-Tooth disease, type 1C	14.3	AD	CMT1C 601098	16p13.13	***LITAF*** *603795*	Lipopolysaccharide-induced TNF-alpha factor	Street et al. (2002, 2003)	
Charcot-Marie-Tooth disease, type 1D	14.4	AD	CMT1D 607678	10q21.3	***EGR2*** *129010*	Early growth response 2 (Krox-20 homolog)	Warner et al. (1998) Street et al. (2003)	allelic to CHN and DSS (group 14)
Charcot-Marie-Tooth disease, type 1E, with deafness	14.5	AD	CMT1E 118300	17p12	***PMP22*** *601097*	Peripheral myelin protein 22	Kovach et al. (1999) Boerkoel et al. (2002)	allelic to CMT1A, HNPP and DSS (group 14),
Hereditary neuropathy with liability to pressure palsies	14.6	AD	HNPP 162500	17p12	***PMP22*** *601097*	Peripheral myelin protein P22	Chance et al. (1993) Nicholson et al. (1994) Mariman et al. (1994)	allelic to CMT1A, CMT1E and DSS (group 14)
Charcot-Marie-Tooth disease, type 1F	14.7	AD	CMT1F 607734	8p21.2	***NEFL*** *162280*	Neurofilament, light polypeptide 68kDa	Jordanova et al. (2003)	allelic to CMTDIG and CMT2E (group 14)
Charcot-Marie Tooth disease, demyelinating type 1G	14.8	AD	CMT1G 618279	8q21.13	***PMP2*** *170715*	Peripheral myelin protein 2	Hong et al. (2016)	
Charcot-Marie-Tooth with congenital vertical talus	14.9	AD	CVT 192950	2q31.1	***HOXD10*** *142984*	Homeobox D10	Shrimpton et al. (2004)	
Slowed nerve conduction velocity	14.10	AD	SNCV 608236	8p23.3	***ARHGEF10*** *608136*	Rho guanine-nucleotide exchange factor-10	De Jonghe et al. (1999) Verhoeven et al. (2003)	

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Neuropathy, hereditary, with or without age-related macular degeneration	14.11	AD	HNARMD 608895	14q32.12	***FBLN5*** *604580*	Fibulin 5	Auer-Grumbach et al. (2011)	
Charcot-Marie-Tooth neuropathy	14.12	AD		6q21.31	***ITPR3*** *147267*	Inositol 1,4,5-triphosphate receptor, type 3	Rönkkö et al. (2020)	
Dominant intermediate (CMTDI)								
Charcot-Marie-Tooth disease, dominant intermediate A	14.13	AD	CMT2GG 606483	10q24.1-q25.1	***GBF1*** *603698*	Golgi-specific brefeldin-A resistance factor 1	Verhoeven et al. (2001) Mendoza-Ferreira et al. (2020)	allelic to Distal hereditary motor neuropathies (group 12)
Charcot-Marie-Tooth disease, dominant intermediate B	14.14	AD	CMTDIB 606482	19p13.2	***DNM2*** *602378*	Dynamin 2	Zuchner et al. (2005)	allelic to CMD related to *DNM2* (group 2), CNM1 (group 3), distal myopathy related to *DNM2* (group 4) and LCCS5 (group 12)
Charcot-Marie-Tooth disease, dominant intermediate C	14.15	AD	CMTDIC 608323	1p35.1	***YARS*** *603623*	Tyrosyl-tRNA synthetase	Jordanova et al. (2003, 2006)	Allelic to MLASA2 (group 16)
Charcot-Marie-Tooth disease, dominant intermediate D	14.16	AD	CMTDID 607791	1q23.3	***MPZ*** *159440*	Myelin protein zero	Mastaglia et al. (1999)	allelic to CMT1B, CMT2I, CMT2J, DSS and CHN (group 14)
Charcot-Marie-Tooth disease, dominant intermediate E	14.17	AD	CMTDIE 614455	14q32.33	***INF2*** *610982*	Inverted formin 2	Boyer et al. (2011)	
Charcot-Marie-Tooth disease, dominant intermediate F	14.18	AD	CMTDIF 615185	3q28.33	***GNB4*** *610863*	Guanine nucleotide-binding protein, beta-4	Soong et al. (2013)	
Charcot-Marie-Tooth disease, dominant intermediate G	14.19	AD	CMTDIG 617882	8p21.2	***NEFL*** *162280*	Neurofilament, light polypeptide 68kDa	Berciano et al. (2015)	allelic to CMT1F and CMT2E (group 14)
Charcot-Marie Tooth disease, intermediate	14.20	AD		1p21.2-p13.3	***C1orf194*** *618682*	Chromosome 1 open reading frame 194	Sun et al. (2019)	
Autosomal recessive (AR-CMT1 or CMT4)								
Charcot-Marie-Tooth disease, type 4A	14.21	AR	CMT4A 214400	8q21.11	***GDAP1*** *606598*	Ganglioside-induced differentiation associated protein1	Ben Othmane et al. (1993) Baxter et al. (2002) Cuesta et al. (2002) Nelis et al. (2002)	allelic to CMT2K and CMTRIA (group 14)
Charcot-Marie-Tooth disease, type 4B1	14.22	AR	CMT4B1 601382	11q21	***MTMR2*** *603557*	Myotubularin-related protein-2	Bolino et al. (1996, 2000) Previtali et al. (2003)	
Charcot-Marie-Tooth disease, type 4B2	14.23	AR	CMT4B2 604563	11p15.4	***SBF2*** *607697*	SET-binding factor 2	Azzedine et al. (2003) Senderek et al. (2004)	
Charcot-Marie-Tooth disease, type 4B3	14.24	AR	CMT4B3 615284	22q13.33	***SBF1*** *603560*	SET-binding factor 1	Nakhro et al. (2013)	
Charcot-Marie-Tooth disease, type 4C	14.25	AR	CMT4C 601596	5q32	***SH3TC2*** *608206*	SH3 domain and tetratricopeptide repeats domain 2	LeGuern et al. (1996) Senderek et al. (2003)	
Charcot-Marie-Tooth disease, type 4D	14.26	AR	CMT4D 601455	8q24.22	***NDRG1*** *605262*	N-myc downstream regulated gene 1	Kalaydjieva et al. (1996, 2000) Hunter et al. (2003)	
Neuropathy, congenital hypomyelinating, 1	14.27	AR	CHN1 605253	10q21.3	***EGR2*** *129010*	Early growth response 2	Warner et al. (1998)	allelic to CMT1D and DSS (group 14)
Neuropathy, congenital hypomyelinating, 2	14.28	AR	CHN2 605253	1q23.3	***MPZ*** *159440*	Myelin protein zero	Warner et al. (1996)	allelic to CMT1B, CMTDID, CMT2I, CMT2J, DSS (group 14)
Charcot-Marie-Tooth disease, type 4F	14.29	AR	CMT4F 614895	19q13.2	***PRX*** *605725*	Periaxin	Delague et al. (2000) Guilbot et al. (2001)	allelic to DSS (group 14)
Neuropathy, hereditary motor and sensory, Russe type	14.30	AR	HMSNR 605285	10q22.1	***HK1*** *142600*	Hexokinase 1	Rogers et al. (2000) Thomas et al. (2001) Hantke et al. (2009) Sevilla et al. (2013)	
Charcot-Marie-Tooth disease, type 4H	14.31	AR	CMT4H 609311	12p11.21	***FGD4*** *611104*	Fyve, RhoGEF and Phdomain-containing protein 4 (Frabin)	De Sandre-Giovannoli et al. (2005) Delague et al. (2007) Stendel et al. (2007)	
Charcot-Marie-Tooth disease, type 4J	14.32	AR	CMT4J 611228	6q21	***FIG4*** *609390*	FIG4, S. Cerevisiae, homolog of	Chow et al. (2007)	allelic to ALS11 (group 12)
Charcot-Marie-Tooth disease, type 4K	14.33	AR	CMT4K 616684	9q34.2	***SURF1*** *185620*	Surfeit 1 (**M**)	Echaniz-Laguna et al. (2013)	
Charcot-Marie Tooth disease	14.34	AR		14q32	***AHNAK2*** *608570*	Ahnak nucleoprotein 2	Tey et al. (2019)	
Charcot-Marie Tooth disease related to *CNTNAP1*	14.35	AR		17q21,2	***CNTNAP1*** *602346*	Contactin-associated protein 1	Freed et al. (2019)	Allelic to LCCS7 (group 12)

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X-linked CMT1								
Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	14.36	XD	CMTX1 302800	Xq13.1	***GJB1*** *304040*	Gap junction protein, beta 1	Bergoffen et al. (1993) Bone et al. (1995)	allelic to DSS (group 14)
Charcot-Marie-Tooth neuropathy, X-linked recessive, 2	14.37	XR	CMTX2 302801	Xp22.2	***?***	?	Ionasecu et al. (1992)	
Charcot-Marie-Tooth neuropathy, X-linked recessive, 3	14.38	XR	CMTX3 302802	Xq26	78 kb ***Chr8 8q24.3 insertion***	78 kb inter-chromosomal insertion (from chr8 - 8q24.3)	Ionasecu et al. (1992) Huttner et al. (2006) Brewer et al. (2016)	
Charcot-Marie-Tooth disease, X-linked 4 (Cowchock syndrome)	14.39	XR	COWCK 310490	Xq26.1	***AIFM1*** *300169*	Apoptosis-inducing factor, mitochondria-associated, 1 (**M**)	Priest et al. (1995) Rinaldi et al. (2012)	Allelic to cerebellar ataxia (group 13) and COXPD6 (group 16)
Charcot-Marie-Tooth disease, X-linked recessive, 5	14.40	XR	CMTX5 311070	Xq22.3	***PRPS1*** *311850*	Phosphoribosyl pyrophosphate synthetase 1	Kim at al (2007)	
Charcot-Marie-Tooth disease, X-linked dominant, 6	14.41	XD	CMTX6 300905	Xp22.11	***PDK3*** *300906*	Pyruvate dehydrogenase kinase, isoenzyme 3	Kennerson et al. (2013)	
Déjerine-Sottas syndrome (DSS or CMT3)								
Déjerine-Sottas hypertrophic neuropathy, dominant	14.42	AD	DSS 145900	17p12	***PMP22*** *601097*	Peripheral myelin protein 22	Roa et al. (1993)	allelic to CMT1A, CMT1E and HNPP (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.43	AD	DSS 145900	1q23.3	***MPZ*** *159440*	Myelin protein zero	Hayasaka et al. (1993)	allelic to CMT1B, CMTD1D, CMT2J, CMT2I and CHN (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.44	AD (digenic)	DSS 145900	10q21.3 and Xq13	***EGR2*** *129010* + ***GJB1*** *304040*	Early growth response 2 + Gap junction protein, beta 1	Chung et al. (2005)	***EGR2:*** allelic to CMT1D and CHN (group 14) ***GJB1***: allelic to CMTX1 (group 14)
Déjerine-Sottas hypertrophic neuropathy, recessive	14.45	AR	DSS 145900	19q13.2	***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 CMT2								
Charcot-Marie-Tooth disease, type 2A1	14.46	AD	CMT2A1 118210	1p36.22	***KIF1B*** *605995*	Kinesin family member 1B	Zhao et al. (2001)	
Charcot-Marie-Tooth disease, axonal, type 2A2A	14.47	AD	CMT2A2A 609260	1p36.22	***MFN2*** *608507*	Mitofusin 2	Ben Othmane et al. (1993) Züchner et al. (2004)	allelic to CMT2A2B and HMSN6A (group 14)
Charcot-Marie-Tooth disease, type 2B	14.48	AD	CMT2B 600882	3q21.3	***RAB7*** *602298*	RAS-associated protein RAB7	Kwon et al. (1995) Pericak-Vance et al. (1997) Kok et al. (2003)	
Hereditary motor and sensory neuropathy, type IIC	14.49	AD	HMSN2C 606071	12q24.11	***TRPV4*** *605427*	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)	allelic to HMN8 and SPSMA (group 12)
Charcot-Marie-Tooth disease, type 2D	14.50	AD	CMT2D 601472	7p14.3	***GARS*** *600287*	Glycyl-tRNA synthetase	Ionasescu et al. (1996) Antonellis et al. (2003)	allelic to HMN5A (group 12)
Charcot-Marie-Tooth disease, type 2E	14.51	AD	CMT2E 607684	8p21.2	***NEFL*** *162280*	Neurofilament, light polypeptide	Birouk et al. (2003) Claramunt et al. (2005)	allelic to CMT1F, CMTDIG (group 14)
Charcot-Marie-Tooth disease, type 2F	14.52	AD, AR	CMT2F 606595	7q11.23	***HSPB1*** *602195*	Heat-shock 27-kD protein 1	Ismailov et al. (2001) Evgrafov et al. (2004) Abati et al. (2021)	allelic to HMN2B (group 14)
Charcot-Marie-Tooth disease, type 2H	14.53	AD	CMT2H 607731	8q13-q23	*?*	?	Barhoumi et al. (2001)	maybe allelic to CMT4A (group 14)
Charcot-Marie-Tooth disease, type 2I	14.54	AD	CMT2I 607677	1q23.3	***MPZ*** *159440*	Myelin protein zero	Auer-Grumbach et al. (2003)	allelic to CMT1B, CMTDID, CMT2J, DSS, CHN (group 14)
Charcot-Marie-Tooth disease, type 2J	14.55	AD	CMT2J 607736	1q23.3	***MPZ*** *159440*	Myelin protein zero	De Jonghe et al. (1999) Chapon et al. (1999)	allelic to CMT1B, CMTDID, CMT2I, DSS, CHN (group 14)

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Charcot-Marie-Tooth disease, type 2K	14.56	AD, AR	CMT2K 607831	8q21.11	***GDAP1*** *606598*	Ganglioside-induced differentiation-associated protein 1	Nelis et al. (2002) Birouk et al. (2003) Claramunt et al. (2005)	allelic to CMT4A and CMTRIA (group 14)
Charcot-Marie-Tooth disease, type 2L	14.57	AD	CMT2L 608673	12q24.23	***HSPB8*** *608014*	Heat-shock 22-kD protein 8	Tang et al. (2004, 2005)	allelic to Rimmed vacuole myopathy and distal myopathy and motor neuropathy (group 4) and HMN2A (group 12)
Charcot-Marie-Tooth disease, type 2N	14.58	AD	CMT2N 613287	16q22.1	***AARS1*** *601065*	Alanyl-tRNA synthetase 1	Latour et al. (2010)	Allelic to dHMN (group 12)
Charcot-Marie-Tooth disease, type 2O	14.59	AD	CMT2O 614228	14q32.31	***DYNC1H1*** *600112*	Dynein, cytoplasmic 1, heavy chain 1	Weedon et al. (2011)	allelic to SMALED1 (group 12)
Charcot-Marie-Tooth disease, type 2P	14.60	AD	CMT2P 614436	9q33.3-q34.1	***LRSAM1*** *610933*	Leucine rich repeat and sterile alpha motif containing 1	Nelis et al. (2004) Guernsey et al. (2010) Weterman et al. (2012) .	
Charcot-Marie-Tooth disease, type 2Q	14.61	AD	CMT2Q 615025	10p14	***DHTKD1*** *614984*	Dehydrogenase E1 and transketolase domain containing 1	Xu et al. (2012)	
Charcot-Marie-Tooth disease, type 2U	14.62	AD	CMT2U 616280	12q13.3	***MARS*** *156560*	Methionyl-tRNA synthetase	Gonzalez et al. (2013)	
Charcot-Marie-Tooth disease, type 2V	14.63	AD	CMT2V 616491	17q21.2	***NAGLU*** *609701*	N-acetyl-alpha-glucosaminidase	Tetreault et al. (2015)	
Charcot-Marie-Tooth disease, type 2W	14.64	AD	CMT2W 616625	5q31.3	***HARS*** *142810*	Histidyl-tRNA synthetase	Vester et al. (2013) Safka-Brozkova et al. (2015)	
Charcot-Marie-Tooth disease, type 2Y	14.65	AD	CMT2Y 616687	9p13.3	***VCP*** *601023*	Valosin-containing protein	Gonsalez et al. (2014)	allelic to Scapuloperoreal myopathy (group 1), IBMPFD1 (groups 4 and 5), ALS14 (group 12)
Charcot-Marie-Tooth disease, type 2Z	14.66	AD	CMT2Z 616688	22q12.2	***MORC2*** *616661*	MORC family CW-type zinc finger 2	Albulym et al. (2015) Sevilla et al. (2016)	
Charcot-Marie-Tooth disease, axonal, type 2CC	14.67	AD	CMT2CC 616924	22q12.2	***NEFH*** *162230*	Neurofilament Protein, Heavy Polypeptide	Rebelo et al. (2016)	allelic to ALS related to NEFH (group 12)
Charcot-Marie-Tooth disease, axonal, type 2DD	14.68	AD	CMT2DD 618036	1p13.1	***ATP1A1*** *182310*	ATPase, Na^+/K^+ transporting, alpha-1 polypeptide	Lassuthova et al. (2018)	
Charcot-Marie-Tooth disease, axonal, type 2FF	14.69	AD	CMD2FF 619519	1q23.2	***CADM3*** *609743*	Cell adhesion molecule 3	Rebelo et al. (2021)	
Hereditary motor and sensory neuropathy, Okinawa type	14.70	AD	HMSNO 604484	3q12.2	***TFG*** *602498*	TRK-fused gene	Takeshima et al. (1997, 1999) Maeda et al. (2007) Ishiura et al. (2012)	allelic to SPG57 (group 15)
Hereditary motor and sensory neuropathy, type VIA with optic atrophy	14.71	AD	HMSN6A 601152	1p36.22	***MFN2*** *608507*	Mitofusin 2	Zuchner et al. (2006)	Allelic to CMT2A2A and CMT2A2B (group 14)
CMT2 related to *KIF5A*	14.72	AD		12q13.3	***KIF5A*** *602821*	Kinesin family member 5A	Liu et al. (2014)	allelic to ALS25 (group 12) and SPG10 (group 15)
Early onset axonal neuropathy with sensory ataxia	14.73	AD	CMT2	1q13.5	***DGAT2*** 606983	Diacylglycerol O-acyltransferase 2	Hong et al. (2016)	
Charcot-Marie-Tooth disease, axonal, related to *BAG3*	14.74	AD		10q26.11	***BAG3*** *603883*	BCL2-associated athanogene 3	Noury et al. (2017)	allelic to MFM6 (group 5) and to CMH1HH (group 10A)
Charcot-Marie-Tooth disease, axonal, related to *JAG1*	14.75	AD	CMT2	20p12.2	***JAG1*** *601920*	Jagged 1	Sullivan et al. (2020)	
Autosomal recessive CMT2								
Charcot-Marie-Tooth disease, axonal, type 2A2B	14.76	AR	CMT2A2B 617087	1p36.22	***MFN2*** *608507*	Mitofusin 2	Polke et al. (2011)	allelic to CMT2A2A and HMSN6A (group 14)
Charcot-Marie-Tooth disease, axonal, type 2B1	14.77	AR	CMT2B1 605588	1q22	***LMNA*** *150330*	Lamin A/C	Bouhouch et al. (1999) De Sandre et al. (2002) Worman and Bonne (2007)	allelic to EDMD2, EDMD3, formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A) [+ several other phenotypes not in this table: FPLD2 #151,660, HGPS #176,670, restrictive dermopathy #275,210, MADA #248,370]

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Charcot-Marie-Tooth disease, axonal, type 2B2	14.78	AR	CMT2B2 605589	19q13.33	***PNKP*** *605610*	Polynucleotide kinase 3-prime phosphatase	Leal et al. (2009) Leal et al. (2018)	Allelic to AOA4 (group 14)
Charcot-Marie-Tooth disease, axonal, type 2EE	14.79	AR	CMT2EE 618400	2p23.3	***MPV17*** *137960*	MPV17, mouse, homolog of (**M**)	Choi et al. (2015) Baumannn et al. (2019)	
Charcot-Marie-Tooth disease, axonal, related to *DNAJB2*	14.80	AR		2q35	***DNAJB2*** *604139*	DnaJ/Hsp40 homolog, subfamily B, member 2	Gess et al. (2014)	allelic to DSMA5 (group 12)
Neuromyotonia and axonal neuropathy, autosomal recessive	14.81	AR	NMAN 137200	5q23.3	***HINT1*** *601314*	Histidine triad nucleotide binding protein 1	Zimon et al. (2012)	Allelic to Distal hereditary motor neuronopathy (group 12)
Charcot-Marie-Tooth disease, recessive intermediate, A	14.82	AR	CMTRIA 608340	8q21.11	***GDAP1*** *606598*	Ganglioside-induced differentiation-associated protein 1	Nelis et al. (2002)	allelic to CMT4A and CMT2K (group 14)
Charcot-Marie-Tooth disease, recessive intermediate, B	14.83	AR	CMTRIB 613641	16q23.1	***KARS*** *601421*	Lysyl-tRNA synthetase	McLaughlin et al. (2010)	
Charcot-Marie-Tooth disease, recessive intermediate, C	14.84	AR	CMTRIC 615376	1p36.31	***PLEKHG5*** *611101*	Pleckstrin homology domain- and RhoGEF domain-containing, family G5	Azzedine et al. (2013) Kim et al. (2013)	allelic to DSMA4 (group 12)
Charcot-Marie-Tooth disease, recessive intermediate, D	14.85	AR	CMTRID 616039	12q24.31	***COX6A1*** *602072*	Cytochrome c oxidase subunit 6a1 (**M**)	Tamiya et al. (2014)	
Charcot-Marie-Tooth disease, type 2R	14.86	AR	CMT2R 615490	4q31.3	***TRIM2*** *614141*	Tripartite motif-containing protein 2	Ylikallio et al. (2013) Pehlivan et al. (2015)	
Charcot-Marie-Tooth disease, type 2S	14.87	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)
Charcot-Marie-Tooth disease, type 2T	14.88	AR	CMT2T 617017	3q25.2	***MME*** *120520*	Membrane metalloendopeptidase	Higuchi et al. (2016)	allelic to SCA43 (group 15)
Charcot-Marie-Tooth disease, type 2X	14.89	AR	CMT2X 616668	15q21.1	***SPG11*** *610844*	SPG11 vesicle trafficking associated Spatacsin	Montecchiani et al. (2015)	allelic to ALS5 (group 12)
Early-onset axonal Charcot-Marie-Tooth with ataxia	14.90	AR	AOA4 616267	19q13.33	***PNKP*** *605610*	Polynucleotide kinase 3′-phosphatase	Pedroso et al. (2015)	allelic to CMT2B2 (group 14)
Charcot-Marie-Tooth disease, axonal	14.91	AR		10q22.1	***SGPL1*** *603729*	Sphingosine-1 phosphate lyase 1	Atkinson et al. (2017)	allelic to Nephrotic Syndrome 14 (# 617575)
Charcot-Marie-Tooth disease, axonal; related to *SCO2*	14.92	AR		22q13.33	***SCO2*** *604272*	Cytochrome c oxidase assembly protein 2	Rebelo et al. (2018)	allelic to CEMCOX1 (group 10A)
Charcot-Marie-Tooth disease, axonal; related to *SACS*	14.93	AR		13q12.12	***SACS*** *604490*	Sacsin	Souza et al. (2018)	allelic to SACS (group 13) and SPAX6 (group 15)
Sorbitol dehydrogenase deficiency with peripheral neuropathy (CMT2 AR)	14.94	AR	SORDD 618912	15q21.1	***SORD*** *182500*	Sorbitol Dehydrogenase	Cortese et al. (2020)	allelic with CMTA Intermediate and dHMN
Charcot-Marie-Tooth disease, axonal	14.95	AR		21q22.3	***MCM3AP*** *603294*	Minichromosome maintenance 3-associated protein	Sedghi et al. (2019)	allelic to PNRIID (group 12)
Charcot-Marie-Tooth disease, axonal	14.96	AR		12q13.3	***B4GALNT1*** *601823*	Beta-1,4-N-Acetyl-galactosaminyl transferase 1	Hong et al. (2021)	allelic to SPG26 (group 15)
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 Hereditary sensory and autonomic neuropathy (HSAN)								
Hereditary sensory and autonomic neuropathy, type IA	14.97	AD	HSAN1A 162400	9q22.31	***SPTLC1*** *605712*	Serine palmitoyl transferase long-chain base subunit 1	Nicholson et al. (1996) Bejaoui et al. (2001) Dawkins et al. (2001)	allelic to Amyotrophic lateral sclerosis (group 12)
Hereditary sensory and autonomic neuropathy, type IB with cough and gastroesophageal reflux	14.98	AD	HSAN1B 608088	3p24-p22	***?***	?	Kok et al. (2004)	
Hereditary sensory and autonomic neuropathy, type IC	14.99	AD	HSAN1C 613640	14q24.3	***SPTLC2*** *605713*	Serine palmitoyl transferase long-chain base subunit 2	Rotthier et al. (2010)	

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Hereditary sensory and autonomic neuropathy, type IIA	14.100	AR	HSAN2A 201300	12p.13.33	***WNK1*** *605232*	Protein kinase, lysine deficient 1	Lafreniere et al. (2004) Shekarabi et al. (2008)	
Hereditary sensory and autonomic neuropath, type IIB	14.101	AR	HSAN2B 613115	5p15.1	***RETREG1*** *(FAM134B)* 613114	Family with sequence similarity 134 member B	Kurth et al. (2009)	
Hereditary sensory and autonomic neuropathy, type IID	14.102	AR	CIP 24300	2q24.3	***SCN9A*** 603415	Sodium channel, voltage-gated alpha subunit	Yuan et al. (2013)	allelic to SFNP (group 14)
Hereditary sensory and autonomic neuropathy type III	14.103	AR	HSAN3 223900	9q31.3	***ELP1*** *603722*	Inhibitor of kappa light polypeptide gene enhanccer in B cells, kinase complex associated protein (IKBKAP)	Blumenfeld et al. (1993) Anderson et al. (2001) Slaugenhaupt et al. (2001)	allelic to familial Dysautonomia (group 16)
Hereditary sensory and autonomic neuropathy type IV	14.104	AR	CIPA 256800	1q23.1	***NTRK1*** 191315	Neurotrophic tyrosine kinase, receptor, type 1	Indo et al. (1996)	
Hereditary sensory and autonomic neuropathy type V	14.105	AR	HSAN5 608654	1p13.1	***NGF*** *162030*	Nerve growth factor (beta polypeptide)	Einarsdottir et al. (2004)	
Hereditary sensory and autonomic neuropathy type VI	14.106	AR	HSAN6 614653	6p12.1	***DST*** 113810	Dystonin	Edvardson et al. (2012)	
Neuropathy, hereditary sensory and autonomic, type VII	14.107	AD	HSAN7 615548	3p22.2	***SCN11A*** 604385	Sodium channel, voltage-gated alpha subunit	Leipold et al. (2013)	allelic to FEPS3 (group 14)
Hereditary sensory and autonomic neuropathy type VIII	14.108	AR	HSAN8 616488	9q34.12	***PRDM12*** *616458*	PR Domain-containing protein 12	Chen et al. (2015)	
Hereditary sensory neuropathy, type ID	14.109	AD	HSN1D 613708	14q22.1	***ATL1*** *606439*	Atlastin GTPase 1	Guelly et al. (2011)	allelic to SPG3A (group 15)
Hereditary sensory neuropathy, type IE	14.110	AD	HSN1E 614116	19p13.2	***DNMT1*** *126375*	DNA methyltransferase 1	Klein et al. (2011)	
Neuropathy, hereditary sensory, type IF	14.111	AD	HSN1F 615632	11q13.1	***ATL3*** 609369	Atlastin GTPase 3	Kornak et al. (2014)	
Hereditary sensory neuropathy, type IIC	14.112	AR	HSN2C 614213	2q37.3	***KIF1A*** *601255*	Kinesin family member 1A	Riviere et al. (2011)	allelic to SPG30 (group 15)
Ataxia, posterior column, with retinitis pigmentosa (PCARP)	14.113	AR	AXPC1 609033	1q32.3	***FLVCR1*** *609144*	Feline leukemia subgroup C receptor 1	Rajadhyaksha et al. (2010)	
Absence of pain, Congenital	14.114	AR		22q11.21	***CLTCL1*** ***601273***	Clathrin, heavy polypepdide-like 1	Nahorski et al. (2015)	
Marsili syndrome (insensitivity to pain, congenital, AD)	14.115	AD	MARSIS 147430	14q11.2	***ZFHX2*** *617828*	Zinc finger homeobox 2	Habib et al. (2018)	
Erythromelalgia, Primary	14.116	AD	SFNP 133020	2q24.3	***SCN9A*** 603415	Sodium channel, voltage-gated alpha subunit	Michiels et al. (2005) Faber et al. (2012)	allelic to CIP (group 14)
Episodic pain syndrome, familial 3	14.117	AD	FEPS3 615552	3p22.2	***SCN11A*** *604385*	Sodium channel, voltage-gated alpha subunit	Zhang et al. (2013) Huang et al. (2014)	allelic to HSAN7 (group 14)
Polyneuropathy with erythromelalgia	14.118	AR		1q25.3	***NMNAT2*** *608701*	Nicotinamide nucleoside adenyltransferase 2	Huppke et al. (2019)	
Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome	14.119	AR	CANVAS 614575	4p14	***RFC1*** *102579*	Replication Factor C Subunit 1	Cortese et al. (2019)	
E. Other complex neuropathy syndromes								
Peripheral neuropathy and agenesis of the corpus callosum (Charlevoix disease)	14.120	AR	ACCPN 218000	15q14	***SLC12A6*** *(KCC3)* *604878*	Solute carrier family 12 (potassium/ chloride transporter), member 6	Casaubon et al. (1996) Howard et al. (2002)	
Peripheral neuropathy and deafness, autosomal dominant	14.121	AD		1p34.3	***GJB3*** *603324*	Gap junction protein, beta 3	Lopez-Bigas et al. (2001)	

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Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy)	14.122	AD	HNA 162100	17q25.3	***SEPT9*** *604061*	Septin 9	Pellegrino et al. (1996) Kuhlenbaumer et al. (2005)	
Giant axonal neuropathy-1	14.123	AR	GAN1 256850	16q23.2	***GAN1*** *605379*	Gigaxonin	Ben Hamida et al. (1997) Bomont et al. (2000)	
Giant axonal neuropathy-2	14.124	AD	GAN2 610100	1q23.2	***DCAF8*** *615820*	DDB1- and CUL4-associated factor 8	Klein et al. (2014)	
Congenital cataracts, facial dysmorphism and neuropathy	14.125	AR	CCFDN 604168	18p23	***CTDP1*** *604927*	C-terminal domain of RNA polymerase II subunit A, phosphatase of, subunit 1	Varon et al. (2003)	
Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis	14.126	AR		14q32.2	***VRK1*** *602168*	Vaccinia related kinase 1	Gonzaga-Jauregui et al. (2013)	Allelic to DSMA and PCH1 (group 12)
Neuropathy, hereditary sensory, with spastic paraplegia	14.127	AR	256840	5p15.2	***CCT5*** *610150*	Chaperonin containing T-complex polypeptide1, subunit 5	Bouhouche et al. (2006)	
Neuronal intranuclear inclusion diseases	14.128	AD	603472	1q21.2	***NOTCH2NLC*** *(NBPF19)* *618025*	Notch2 N-terminal-like protein	Tian et al. (2019) Sone et al. (2019) Ishiura et al. (2019)	
Hereditary peripheral neuropathy (CMT ?)	14.129	AD		17q25.1	***SLC9A3R1*** *(EBP50)* *604990*	Solute carrier family 9, member 3, regulator 1	Song et al. (2020)	
Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities	14.130	AR, AD	NEDMILG 619091	18q21.31	***NARS1*** *108410*	Asparaginyl-tRNA Synthetase 1	Manole et al. (2020)	
Group 16. OTHER NEUROMUSCULAR DISORDERS								
Torsion dystonia 1, early onset	16.1	AD	DYT1 128100	9q34.11	***TOR1A*** *605204*	Torsin 1A	Ozelius et al. (1997) Ikeuchi et al. (1999)	
Myoclonic dystonia 11	16.2	AD	DYT11 159900	7q21.3	***SGCE*** *604149*	Sarcoglycan, Epsilon	Klein et al. (2000) Zimprich et al. (2001) Tezenas du Montcel et al. (2006)	
Hereditary neuropathy, sensory and autonomic (Riley-Day syndrome)	16.3	AR	HSAN3 223900	9q31.3	***ELP1*** *603722*	Inhibitor of kappa Light polypeptide gene enhaner in B cells, kinase complex associated protein	Blumenfeld et al. (1993) Anderson et al. (2001) Slaugenhaupt et al. (2001)	allelic to HSNA3 (group 14)
Hereditary amyloidosis transthyretin-related	16.4	AD	105210	18q12.1	***TTR*** *176300*	Transthyretin	Costa et al. (1978) Tawara et al. (1983) Saraiva et al. (1995)	
Fibrosis of extraocular muscles, congenital, 1	16.5	AD	CFEOM1 CFEOM3B 135700	12q12	***KIF21A*** *608283*	Kinesin family member 21A	Engle et al. (1994) Yamada et al. (2003) Tiab et al. (2004)	
Fibrosis of extraocular muscles, congenital, 2	16.6	AD	CFEOM2 602078	11q13.4	***PHOX2A*** *602753*	Aristaless homeobox, drosophila, homolg of, (ARIX)	Wang et al. (1998) Nakano et al. (2001)	
Fibrosis of extraocular muscles, congenital, 3A, with or without extraocular involvement	16.7	AD	CFEOM3A 600638	16q24.3	***TUBB3*** *602661*	Tubulin, beta-3	Doherty et al. (1999) Tischfield et al. (2010)	
Fibrosis of extraocular muscles, congenital, 5	16.8	AR	CFEOM5 66219	4q25	***COL25A1*** *610004*	Collagen, type XXV, alpha-1	Shinwari et al. (2015)	
Arthrogryposis, distal, type 1A	16.9	AD	DA1A 108120	9p13.3	***TPM2*** *190990*	Tropomyosin-2 (beta)	Sung et al. (2003)	allelic to NEM4 and CAPM2 (group 3), DA2B (group 16)
Arthrogryposis, distal, type 1B	16.10	AD	DA1B 614335	12q23.2	***MYBPC1*** *160794*	Myosin-binding proteinC, slow type	Gurnett et al. (2010)	Allelic to LCCS4 (group 12)
Arthrogryposis, distal, type 2A, Freeman-Sheldon syndrome	16.11	AD	DA2A 193700	17p13.1	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al. (2006)	Allelic to DA2B (group 16)

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Arthrogryposis, distal, type 2B1, Sheldon-Hall syndrome	16.12	AD	DA2B1 601680	11p15.5	***TNNI2*** *191043*	Troponin I, fast skeletal	Sung et al. (2003a) Kimber et al. (2006)	
Arthrogryposis, distal, type 2B2	16.13	AD	DA2B2 618435	11p15.5	***TNNT3*** *600692*	Troponin T3, fast skeletal	Sung et al. (2003)	Allelic to Nemalin myopathy with distal arthrogryposis (group 3)
Arthrogryposis, distal, type 2B3	16.14	AD	DA2B3 618436	17p13.1	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al. (2006)	Allelic to DA2A (group 16)
Arthrogryposis, distal, type 2B4	16.15	AD	DA2B4 108120	9p13.3	***TPM2*** *190990*	Tropomyosin-2 (beta)	Tajsharghi et al. (2007) Ochala et al. (2007)	allelic NEM4 and CAPM2 (group 3), DA1A (group 16)
Arthrogryposis, distal, type 3	16.16	AD	DA3 114300	18p11.22-p11.21	***PIEZO2*** *613629*	Piezo-type mechanosensitive ion channe component 2	McMillin et al. (2014)	Allelic to DA5 (group 16)
Arthrogryposis, distal, type 5	16.17	AD	DA5 108145	18p11.22-p11.21	***PIEZO2*** *613629*	Piezo-type mechanosensitive ion channe component 2	Coste et al. (2013)	Allelic to DA3 (group 16)
Arthrogryposis, distal, type 5D	16.18	AR	DA5D 615065	2q37.1	***ECEL1*** *605896*	Endothelin-converting enzyme like 1	McMillin et al. (2013)	
Atrthrogryposis, Distal, type 7 (Trismus-pseudocamptodactyly syndrome)	16.19	AD	DA7 158300	17p13.1	***MYH8*** *160741*	Myosin heavy chain, 8, skeletal muscle, perinatal	Veugelers et al. (2004) Toydemir et al. (2006b)	
Arthrogryposis, distal, type 10	16.20	AD	DA10 187370	2q31.3-q32.1	*?*	?	Stevenson et al. (2006a) Stevenson et al. (2006b)	
Arthrogryposis multiplex congenita with nesprin-1 defect	16.21	AR	AMCM 618484	6q25.2	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (Nesprin-1)	Attali et al. (2009)	allelic to EDMD4 (group 1), DCM related to *SYNE1* (group 10A) and SCAR8 (group 13)
Arthrogryposis and BICD2-related neuromuscular disease	16.22	AD		9q22.31	***BICD2*** *609797*	Bicaudal D homolog 2 (Drosophila)	Storbeck et al. (2017)	Allelic to SMALED2A and SMALED2B (group 12)
Arthrogryposis and muscular dysplasia	16.23	AD		7q31.2	***MET*** *164860*	MET protooncogene	Zhou et al. (2019)	
Arthrogryposis related to *ASCC1*	16.24	AR		10q22.1	***ASCC1*** *614215*	Activating signal cointegrator 1 complex subunit 1	Böhm et al. (2019)	allelic to SMABF2 (group 12)
Neurodevelopmental disorder with microcephaly, arthrogryposis and structural brain anomalies	16.25	AR	NEDMABA 618622	2q21.1	***SMPD4*** *610457*	Sphingomyelin phosphodiesterase 4, neutral membrane	Magini et al. (2019)	
Fetal akinesia deformation sequence 1	16.26	AR	FADS1 208150	9q31.3	***MUSK*** *601296*	Muscle-specific receptor tyrosine kinase	Tan-Sindhunata et al. (2015)	allelic to CMS9 (group 11)
Fetal akinesia deformation sequence 3	16.27	AR	FADS3 618389	4p16.3	***DOK7*** *610285*	Downstream of tyrosin kinase 7	Vogt et al. (2009)	allelic to CMS10 (group 11)
Fetal akinesia deformation sequence 2	16.28	AR	FADS2 618388	11p11.2	***RAPSN*** *601592*	Receptor-associated protein of the synapse, 43 kD (Rapsyn)	Vogt et al. (2008)	allelic to CMS11 (group 11)
Fetal akinesia deformation sequence 4	16.29	AR	FADS4 618393	17p13.2	***NUP88*** *602552*	Nucleoporin 88kD	Bonnin et al. (2018)	
Fetal akinesia deformation sequence related to *RYR1*	16.30	AR		19q13.2	***RYR1*** *180901*	Ryanodine receptor	Romero et al. (2003) McKie et al. (2014) Alkhunaizi et al. (2019)	allelic to CMD related to *RYR1* (group 2), CFTD, CNM related to *RYR1,* CCD, CNMDU1, minicore myopathy with external ophthalmoplegia, DuCD (group 3), Late onset axial myopathy (group 5), MHS1 (group 8)
Fetal akinesia deformation related to *AGRN*	16.31	AR		1p36.33	***AGRN*** *103320*	Agrin	Geremek et al. (2020)	Allelic to CMS8 (group 11)
Severe foetal hypokinesia related to *SCN4A*	16.32	AR		17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Zaharieva et al. (2019)	allelic to Congenital Myopathy related to *SCN4A* (group 3), HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1	16.33	AD	PEOA1 157640	15q26.1	***POLG*** *174763*	Polymerase, DNA, gamma (**M**)	Van Goethem et al. (2001)	allelic to SANDO (group 13), MTDPS4A, MTDPS4B, PEOB1 (group 16)

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Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2	16.34	AD	PEOA2 609283	4q35	***SLC25A4*** *103220*	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 (**M**)	Kaukonen et al. (2000)	Allelic to MTDPS12A and MTDPS12B (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3	16.35	AD	PEOA3 609286	10q24.31	***TWNK*** *606075*	Twinkle, mtDNA helicase (**M**)	Suomalinen et al. (1997) Spelbrink et al. (2001)	allelic to IOSCA (group 13)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4	16.36	AD	PEOA4 610131	17q23.3	***POLG2*** *604983*	Polymerase DNA, gamma 2 (**M**)	Longley et al. (2006)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5	16.37	AD	PEOA5 613077	8q22.3	***RRM2B*** *604712*	Ribonucleotide reductase, M2 B (**M**)	Tyynismaa et al. (2009)	allelic to MTDP8A (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6	16.38	AD	PEOA6 615156	10q21.3	***DNA2*** *601810*	DNA replication helicase 2 (**M**)	Ronchi et al. (2013)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 1	16.39	AR	PEOB1 258450	15q26.1	***POLG*** *174763*	Polymerase, DNA, gamma (**M**)	Deschauer et al. (2007)	allelic to SANDO (group 13), MTDPS4A, MTDPS4B, PEOA1 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	16.40	AR	PEOB2 616479	2p25.3	***RNASEH1*** *604123*	Ribonuclease H1 (**M**)	Reyes et al. (2015)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3	16.41	AR	PEOB3 617069	16q21	***TK2*** *188250*	Thymidine kinase, mitochondrial (**M**)	Tyynismaa et al. (2012)	allelic to MTDPS2 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4	16.42	AR	PEOB4 617070	2p13.1	***DGUOK*** *601465*	Deoxyguanosine kinase (**M**)	Ronchi et al. (2012)	Allelic to MTDPS3 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5	16.43	AR	PEOB5 618098	17p11.2	***TOP3A*** *601243*	DNA topoisomerase III	Nicholls et al. (2018)	
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	16.44	AR	MTDPS1 603041	22q13.33	***TYMP*** *131222*	Thymidine phosphorylase	Nishino et al. (1999)	
Mitochondrial DNA depletion syndrome 2 (myopathic type)	16.45	AR	MTDPS2 609560	16q21	***TK2*** *188250*	Thymidine kinase, mitochondrial (**M**)	Saada et al. (2001)	allelic to PEOB3 (group 16)
Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	16.46	AR	MTDPS3 251880	2p13.1	***DGUOK*** *601465*	Deoxyguanosine kinase (**M**)	Mandel et al. (2001)	Allelic to Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4 (group 16)
Mitochondrial DNA depletion syndrome 4A (Alpers type)	16.47	AR	MTDPS4A 203700	15q26.1	***POLG*** *174763*	Polymerase, DNA, gamma (**M**)	Naviaux and Nguyen (2004)	Allelic to SANDO (group 13), MTDPS4B, PEOA1 and PEOB1 (group 16)
Mitochondrial DNA depletion syndrome 4B (MNGIE type)	16.48	AR	MTDPS4B 613662	15q26.1	***POLG*** *174763*	Polymerase, DNA, gamma (**M**)	Van Goethem et al. (2003)	Allelic to SANDO (group 13), MTDPS4A, PEOA1 and PEOB1 (group 16)

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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)
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	16.49	AR	MTDPS5 612073	13q14.2	***SUCLA2*** *603921*	Succinate-CoA ligase, ADP-forming, beta subunit (**M**)	Elpeleg et al. (2005)	
Mitochondrial DNA depletion syndrome 8A (Encephalomyopathic type with renal tubulopathy)	16.50	AR	MTDP8A 612075	8q22.3	***RRM2B*** *604712*	Ribonucleotide reductase, M2B (**M**)	Bourdon et al. (2007)	allelic to PEOA5 and MTDPS8B (group 16)
Mitochondrial DNA depletion syndrome 8B (MNGIE type)	16.51	AR	MTDP8B 612075	8q22.3	***RRM2B*** *604712*	Ribonucleotide reductase, M2B (**M**)	Bourdon et al. (2007)	allelic to PEOA5 and MTDPS8A (group 16)
Mitochondrial DNA depletion syndrome 9 (Encephalomyopathic type withmethylmalonic aciduria)	16.52	AR	MTDPS9 245400	2p11.2	***SUCLG1*** *611224*	Succinate-CoA ligase, alpha subunit (**M**)	Ostergaard et al. (2007)	
Mitochondrial DNA depletion syndrome 11	16.53	AR	MTDPS11 615084	20p11.23	***MGME1*** *615076*	Mitochondrial genome maintenance exonuclease 1 (**M**)	Kornblum et al. (2013)	
Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type)	16.54	AD	MTDPS12A 617184	4q35.1	***SLC25A4*** *103220*	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 (**M**)	Thompson et al. (2016)	Allelic to PEOA2 and MTDPS12B (group 16)
Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type)	16.55	AR	MTDPS12B 615418	4q35.1	***SLC25A4*** *103220*	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 (**M**)	Palmieri et al. (2005)	Allelic to PEOA2 and MTDPS12A (group 16)
Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	16.56	AR	MTDPS13 615471	6q16.1-q16.2	***FBXL4*** *605654*	F-box and leucine-rich repeat protein 4	Bonnen et al. (2013) Gai et al. (2013)	
Mitochondrial DNA depletion syndrome 14 (cardioencephalomyopathic type)	16.57	AR	MTDPS14 616896	3q29	***OPA1*** *605290*	OPA1 protein (M)	Spiegel et al. (2016)	Allelic to Progressive external ophthalmoplegia with optic atrophy, optic atrophy 1 with deafness (group 16)
Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression	16.58	AR	MECREN 618416	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	16.59	AD	125250	3q29	***OPA1*** *605290*	OPA1 protein (**M**)	Amati-Bonneau et al. (2008) Hudson et al. (2008)	Allelic to MTDPS14 (group 16)
Myopathy, lactic acidosis, and sideroblastic anemia 1	16.60	AR	MLASA1 600462	12q24.33	***PUS1*** *608109*	Pseudourine synthase 1 (**M**)	Bykhovskaya et al. (2004) Fernandez-Vizarra (2007)	
Myopathy, lactic acidosis, and sideroblastic anemia-2	16.61	AR	MLASA2 613561	12p11.21	***YARS2*** *610957*	Tyrosyl-tRNA synthetase 2 (**M**)	Riley et al. (2010)	Allelic to CMTDIC (group 14)
Isolated mitochondrial myopathy	16.62	AD	IMMD 616209	22q11.23	***CHCHD10*** *615903*	Coiled-coil-helix-coiled-coil-helix domain containing 10 (**M**)	Ajroud-Driss et al. (2014)	allelic to FTDALS2 and SMAJ (group 12)
Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis	16.63	AD	POIKTMP 615704	11q12.1	***FAM111B*** *615584*	Family with sequence similarity 111, member B	Mercier et al. (2015)	
Combined oxidative phosphorylation deficiency 6;	16.64	XL	COXPD6 300816	Xq26.1	***AIFM1*** *300169*	Apoptosis-inducing factor, mitochondria-associated, 1 (**M**)	Morton et al. (2017)	Allelic to cerebellar ataxia (group 13) and COCK (group 14)
Mitochondrial myopathy with severe neurological manifestations	16.65	AR		8q24.13	***TMEM65*** *616609*	Transmembrane protein 65	Nasli et al. (2017)	
Mitochondrial complex IV deficiency	16.66	AR	220110	2q33.3	***FASTKD2*** *612322*	Fast kinase domains 2 (**M**)	Yoo et al. (2017)	
Mitochondrial complex IV deficiency	16.67	AR	220110	*16p11.2*	***COX6A2*** *6020009*	Cytochrome c-oxidase, subunit 6A2 (**M**)	Inoue et al. (2019)	

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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)
Infantile-onset multisystem disease with progressive muscle weakness	16.68	AR	IMNEPD 616263	17q23.1	***PTRH2*** *608625*	Peptidyl-tRNA hydrolase 2	Hu et al. (2014)	
Dyskinetic cerebral palsy, partial agenesis of the corpus callosum and mitochondrial myopathy	16.69	AR		3p25.1	***MRPS25*** *611987*	Mitochondrial ribosomal protein S25 (**M**)	Bugiardini et al. (2019)	
Myopathy, mitochondrial and cerebellar ataxia (also listed in group 13)	16.70	AR	MMYAT 617675	1q22	***MSTO1*** *617619*	Misato 1, Mitochondrial Distribution and Morphology Regulator (**M**)	Nasca et al. (2017) Ardicli et al. (2019) Donkervoort et al. (2019)	allelic to CMD related to *MSTO1* (group 2)
Early onset of mitochondrial myopathy	16.71	AR		17q13.3	***TIMM22*** *607251*	Translocase of inner mitochondrial membrane 22 (**M**)	Pacheu-Grau et al. (2018)	
Coenzyme Q10 deficiency 1	16.72	AR	COQ10D1 607426	4q21.23	***COQ2*** *609825*	Coenzyme Q2 (**M**)	Quinzii et al. (2006)	
Autosomal recessive spinocerebellar ataxia, 9 with ubiquinone deficiency (also listed in group 13)	16.73	AR	SCAR9 (COQ10D4) 612016	1q42.13	***ADCK3*** *(COQ8A, CABC1) 606980*	Coenzyme Q8A (**M**)	Lagier-Tourenne et al. (2008) Mollet et al. (2008)	
Coenzyme Q10 deficiency 5	16.74	AR	COQ10D5 614654	16q21	***COQ9*** *612837*	Coenzyme Q9 (**M**)	Rahman et al. (2001) Duncan et al. (2009)	
Coenzyme Q10 deficiency 6	16.75	AR	COQ10D6 614650	14q24.3	***COQ6*** *614647*	Coenzyme Q6 (**M**)	Heeringa et al. (2011)	
Coenzyme Q10 deficiency 7	16.76	AR	COQ10D7 607426	9q34.11	***COQ4*** *612898*	Coenzyme Q4 (**M**)	Brea-Calvo et al. (2015)	
Coenzyme Q10 deficiency 8	16.77	AR	COQ10D8 616733	16p12.3	***COQ7*** *601683*	Coenzyme Q7 (**M**)	Freyer et al. (2015)	
Sensory motor axonal neuropathy and myopathy	16.78	AR		19p13.2	***FDX2*** *614585*	Ferredoxin (**M**)	Gurgel-Giannetti et al. (2019)	
Spectraplakinopathy	16.79	AR		1p34.3	***MACF1*** *608271*	Microtubule-actin cross-linking factor 1	Kang et al. (2020)	allelic to LIS9 (#618325)
Mitochondrial complex I deficiency, nuclear type 29	16.80	AR	MC1DN29 618250	11q14.1	***TMEM126B*** *615533*	Transmembrane protein 126B (**M**)	Alston et al. (2016) Sanchez-Caballero et al. (2016)	
Mitochondrial myopathy with lactic acidosis, cognitive impairment and autistic features	16.81	XL		Xp22.11	***APOO*** *300753*	Apolipoprotein O	Beninca et al. (2021)	
Segmental amyoplasia with Distal Arthrogryposis	16.82	AD	DA1C 619110	16p11.2	***MYLPF*** *617378*	Myosin Light Chain Phosphorylatable Fast Skeletal Muscle	Chong et al. (2020)	
Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies	16.83	AR	MYODRIF 618975	11p15.1	***MYOD1*** *159970*	Myogenic Differentiation Antigen 1	Watson et al. (2016)	
Myopathy, mitochondrial progressive, with congenital cataract and developmental delay	16.84	AR	MPMCD 613076	16p13.3	***GFER*** *600924*	Growth Factor ERV1-Like	Di Fonzo et al. (2009)	
Combined oxidative phosphorylation deficiency	16.85	AR	COXPD48 619012	3q11.2	***NSUN3*** *617491*	NOP2/SUN RNA Methyltransferase Family Member 3	Van Haute et al. (2016)	
Mitochondrial neurogastrointestinal encephalomyopathy	16.86	AR		17q12	***LIG3*** *600940*	Ligase III DNA ATP-Dependent	Bonora et al. (2021)	
Mitochondrial complex IV deficiency	16.87	AR	MC4DN22 619355	14q24.2	***COX16*** *618064*	Cytochrome c Oxidase Assembly Factor 16	Wintjes et al. (2021)	

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