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The 2024 version of the gene table of neuromuscular disorders (nuclear genome)

Louise Benarroch^a, Gisèle Bonne^{a,*}, François Rivier^b, Dalil Hamroun^c

^aSorbonne Université, INSERM, Institut de Myologie, Centre de Recherche en Myologie, Paris, France

^bNeuropédiatrie & CR Maladies Neuromusculaires, CHU de Montpellier, U1046 INSERM, UMR9214 CNRS, Université de Montpellier, France

^cCHRU de Montpellier, Direction de la Recherche et de l'Innovation, Hôpital La Colombière, Montpellier, France

General features

This table used to be published annually in the December issue since 2010, but is now published in the January 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 table³

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 <https://www.musclegenetable.fr>. It is designed to cope with the complexity described above. In this version the data are cross-referenced and linked to *PubMed* and to major databases related to molecular medicine (*Leiden Muscular Dystrophy*, *OMIM*, *NCBI*, *GenAtlas*, *Orphanet*, *GeneCards*). It contains several query tools allowing one to perform a variety of interrogations.

* Corresponding author. Sorbonne Université, INSERM, Centre de Recherche en Myologie, Paris, France.

E-mail address: g.bonne@institut-myologie.org (G. Bonne).

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.)

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 2024 printed version of the gene table (pages xx to xxx of this issue)

There are 27 new items, marked by background shading. Altogether they comprise **16 additional genes**, **11 additional phenotypic variants** caused by a gene already listed in the 2023 version. **One locus** without identified gene, on chromosome 19p13, has been suppressed (formerly item #4.16 in the Gene Table 2023), as the original family was solved with the identification of a mutation in the *DNAJB6* gene on chromosome 7q36.

The new key references of the printed version of the table are listed on pages xxx–xxx 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 neuromuscular disorders, in order to allow further time for the neuromuscular community to fully validate these proposed nomenclatures.

Citation of the gene table

- Printed version: Benarroch L, Bonne G, Rivier F, Hamroun D. The 2024 version of the gene table of neuromuscular disorders. *Neuromuscul Disord.* xx (x), xx–xxx.
- Online version: Gene Table of Neuromuscular Disorders: <https://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 for their careful review of entries: Tanya Stojkovic for groups 12 and 14, Jim Dowling for groups 2 and 3 and Volker Straub for group 1.

We acknowledge the help of Myobase, a bibliographic alert system of the AFM (Association 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 2024 printed version of the gene table

16 genes added:

HMGCR (item # 1.45)
BET1 (item # 2.53)
TRAPPC2L (item # 2.53)
MYMX (item # 3.56)
FILIP1 (item # 5.17)
ZC4H2 (item # 5.51)
KLHL24 (item # 10.31)
VEZFI (item # 10.79)
GET3 (item # 10.89)
CAP2 (item # 10.90)
EMILIN1 (item # 12.33)
ATPIA1 (item # 14.14)
SARS1 (item # 14.27)
DRP2 (item # 14.49)
DHX9 (item # 14.86)
ADAMTS15 (item # 16.22)

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

DTNA (item # 1.61)
DPM3 (item # 2.39)
ACTA1 (item # 3.3)
MYBPC1 (item # 3.44)
CACNA1S (item # 3.46)
SCN4A (item # 3.49)
REEP1 (item # 12.18)
SORD (item # 12.20)
COQ7 (item # 12.21)
LRP12 (item # 12.77)
CRYAB (item # 14.85)

33 new key references

Gene table of monogenic neuromuscular disorders (nuclear genome only)

Vol. 33 No. 1, January 2024

A computerized version of the table is freely accessible at <https://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 | XLR | 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) Hodgson et al (1986) Romeo et al (1988) Bione et al (1994, 1995) Klauck et al (1995) Nigro et al (1995) | allelic to CMD3B (group10A) |
| Emery-Dreifuss muscular dystrophy, X-linked, type 1 | 1.2 | XLR | EDMD1 310300 | Xq28 | EMD 300384 | Emerin | Bonne et al (1999) Worman and Bonne (2007) | Allelic to EDMD3 (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) |
| Emery-Dreifuss muscular dystrophy 2, autosomal dominant | 1.3 | AD | EDMD2 181350 | 1q22 | LMNA 150330 | Lamin A/C | Raffaele di Barletta et al (2000) Worman and Bonne (2007) | [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370] |
| Emery-Dreifuss muscular dystrophy 3, autosomal recessive | 1.4 | AR | EDMD3 616516 | 1q22 | LMNA 150330 | Lamin A/C | Zhang et al (2007) | allelic to EDMD2 (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) |
| Emery-Dreifuss muscular dystrophy 4, autosomal dominant | 1.5 | AD | EDMD4 612998 | 6q25.2 | SYNE1 608441 | Spectrin repeat containing, nuclear envelope 1 (nesprin-1) | Zhang et al (2007) | [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370] |
| Emery-Dreifuss muscular dystrophy 5, autosomal dominant | 1.6 | AD | EDMD5 612999 | 14q23.2 | SYNE2 608442 | Spectrin repeat containing, nuclear envelope 2 (nesprin-2) | Zhang et al (2007) | allelic to DCM related to SYNE1 (group 10A), SCAR8 (group 13), AMCM (group 16) |
| Emery-Dreifuss muscular dystrophy 6, X-linked | 1.7 | XLR | 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 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 | TORIAIPI (=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* 606009) | Structural maintenance of chromosomes flexible hinge domain containing 1 | de Greef et al (2010) Sacconi et al (2012) Lemmers et al (2012) Sacconi et al (2013) | allelic to Bosma Arhinia Microphthalmia Syndrome; BAMS (#603457) |
| 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) | |
| Lipodystrophy, congenital generalized, type 4 | 1.13 | AD | CGL4 613327 | 17q21.2 | CAVIN1 603198 | Caveolae-associated protein 1 | Hayashi et al (2009) | |
| muscular dystrophy with gnathodiaphyseal dysplasia | 1.14 | AR | | 11p14.3 | ANOS (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 | 1.15 | AR | MDHLO 619518 | 1q42.3 | GGPS1 606982 | Geranylgeranyl Diphosphate Synthase 1 | Foley et al (2020) | |
| Limb girdle muscular dystrophies, dominant | | | | | | | | |
| Muscular dystrophy, limb-girdle, autosomal dominant 1 | 1.16 | AD | LGMD1 (LGMD1E) 603511 | 7q36.3 | DNAJB6 611332 | Hsp40 homologue, subfamily B, number 6 | Speer et al (1999), Sarparanta et al (2012) Harms et al (2012) | allelic to Rimmed vacuole distal myopathy and distal myopathy (group 4) |
| Muscular dystrophy, limb-girdle, autosomal dominant 2 | 1.17 | AD | LGMD2 (LGMD1F) 608423 | 7q32.1 | TNPO3 610032 | Transportin 3 | Palenzuela et al (2003) Melià et al (2013) Torella et al (2013) | |
| Muscular dystrophy, limb-girdle, autosomal dominant 3 | 1.18 | AD | LGMD3 (LGMD1G) 609115 | 4q21.22 | HNRNPDL 607137 | Heterogeneous nuclear ribonucleoprotein D-like | Starling et al (2005) Vieira et al (2014) | |
| Muscular dystrophy, limb-girdle, autosomal dominant 4 | 1.19 | AD | LGMD4 (LGMD1I) 618129 | 15q15.1 | CAPN3 114240 | Calpain-3 | Vissing et al (2016) Martinez-Thompson et al (2018) | allelic to LGMDR1 (group 1) |
| Muscular dystrophy, limb-girdle, autosomal dominant 5 | 1.20 | AD | BTHLM1 (LGMD5) 158810 | 21q22.3 | COL6A1 120220 | Collagen type VI subunit alpha 1 | Jöbsis et al (1996) | allelic to LGMDR22 (group 1), UCMD1 and BTHLM1 (group 2) |
| Muscular dystrophy, limb-girdle, autosomal dominant 5 | 1.21 | AD | BTHLM1 (LGMD5) 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) |
| Muscular dystrophy, limb-girdle, autosomal dominant 5 | 1.22 | AD | BTHLM1 (LGMD5) 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) |
| Rippling muscle disease 2 | 1.23 | 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) |
| LGMD related to CACNA1S, late-onset | 1.24 | AD | | 1q32.1 | CACNA1S 114208 | Calcium channel, voltage-dependent, L type, alpha 1S subunit | López-Hernández et al (2021) | Allelic to CMYP18 (group 3), HOKPP1 (group 7) and MHS5 (group 8) |
| Limb girdle muscular dystrophies, recessive | | | | | | | | |
| Muscular dystrophy, limb-girdle, autosomal recessive 1 | 1.25 | 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 LGMD4 (group 1) |

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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) |
|---|-------------------------|-------------|---|--------------|--|--|---|---|
| Muscular dystrophy, limb-girdle, autosomal recessive 2 | 1.26 | AR | LGMDR2 (LGMD2B) 253601 | 2p13.2 | <i>DYSF</i> 603009 | Dysferlin | Bashir et al (1994) Bashir et al (1998) Liu et al (1998) | allelic to MMD1 (group 4) |
| Muscular dystrophy, limb-girdle, autosomal recessive 3 | 1.27 | AR | LGMDR3 (LGMD2D) 608099 | 17q21.33 | <i>SGCA</i> 600119 | Alpha-sarcoglycan | Roberds et al (1994) Piccolo et al (1995) Passos-Bueno et al (1995) Ljunggren et al (1995) Carrié et al (1997) | |
| Muscular dystrophy, limb-girdle, autosomal recessive 4 | 1.28 | AR | LGMDR4 (LGMD2E) 604286 | 4q12 | <i>SGCB</i> 600900 | Beta-sarcoglycan | Lim et al (1995) Bönnemann et al (1995) Bönnemann et al (1996) | |
| Muscular dystrophy, limb-girdle, autosomal recessive 5 | 1.29 | AR | LGMDR5 (LGMD2C) 253700 | 13q12.12 | <i>SGCG</i> 608896 | Gamma-sarcoglycan | Ben Othmane et al. (1992) Azibi et al. (1993) Noguchi et al (1995) McNally et al (1996) Piccolo et al (1996) Passos-Bueno et al (1996) | |
| Muscular dystrophy, limb-girdle, autosomal recessive 6 | 1.30 | AR | LGMDR6 (LGMD2F) 601287 | 5q33.3-q33.3 | <i>SGCD</i> 601411 | Delta-sarcoglycan | Nigro et al (1996) Moreira et al (1997) Moreira et al (2000) | allelic to CMD1L (group 10A) |
| Muscular dystrophy, limb-girdle, autosomal recessive 7 | 1.31 | AR | LGMDR7 (LGMD2G) 601954 | 17q12 | <i>TCAP</i> 604488 | Titin-cap (telethonin) | | allelic to CMD related to <i>TCAP</i> (group 2), CMH25 (group 10A), CMD1N (group 10A) |
| Muscular dystrophy, limb-girdle, autosomal recessive 8 | 1.32 | AR | LGMDR8 (LGMD2H) 254110 | 9q33.1 | <i>TRIM32</i> 602290 | Tripartite motif-containing 32 | Weiler et al (1998) Frosk et al (2002) | allelic to sarcotubular myopathy (group 3) |
| Muscular dystrophy, limb-girdle, autosomal recessive 10 | 1.33 | AR | LGMDR10 (LGMD2J) 608807 | 2q31.2 | <i>TTN</i> 188840 | Titin | Hackman et al (2003) | Allelic to CNM related to <i>TTN</i> , MmD related to <i>TTN</i> and CMYP5 (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10), CMD1G (group 10), LCCS related to <i>TTN</i> (group 12) |
| Muscular dystrophy, limb-girdle, autosomal recessive 12 | 1.34 | AR | LGMDR12 (LGMD2L) 611307 | 11p14.3 | <i>ANOS</i> (<i>TMEM16E</i>) 608662 | Anoctamin 5 | Jarry et al (2007) Bolduc et al (2008, 2010) Hicks et al (2011) Gundesli et al (2010) | allelic to Muscular dystrophy with gnathodiaphyseal dysplasia (group 1) and MMD3 (group 4) |
| Muscular dystrophy, limb-girdle, autosomal recessive 17 | 1.35 | AR | LGMDR17 (LGMD2Q) 613723 | 8q24.3 | <i>PLEC</i> 601282 | Plectin | | allelic to LGMD with ophthalmoplegia (group 1), EBSMD (group 5), and Myasthenic syndrome related to <i>PLEC</i> (group 11) |
| Muscular dystrophy, limb-girdle, autosomal recessive 18 | 1.36 | AR | LGMDR18 (LGMD2S) 615356 | 4q35.1 | <i>TRAPPC11</i> 614138 | Trafficking protein particle complex 11 | Bögershausen et al (2013) | allelic to CMD related to <i>TRAPPC11</i> (group 2) |
| Muscular dystrophy, limb-girdle, autosomal recessive 21 | 1.37 | AR | LGMDR21 (LGMD2Z) 617232 | 3q13.33 | <i>POGLUT1</i> 615618 | Protein O-Glucosyltransferase 1 | Servian-Morilla et al (2016) | |
| Muscular dystrophy, limb-girdle, autosomal recessive 22 | 1.38 | AR | UCMD1 (LGMD22) 254090 | 21q22.3 | <i>COL6A1</i> 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) |
| Muscular dystrophy, limb-girdle, autosomal recessive 22 | 1.39 | AR | UCMD1 (LGMD22) 254090 | 21q22.3 | <i>COL6A2</i> 120240 | Collagen type VI subunit alpha 2 | Jokela et al (2019) | allelic to LGMDD5 (group 1), UCMD1, BTHLM1 and congenital myosclerosis (group 2) |
| Muscular dystrophy, limb-girdle, autosomal recessive 22 | 1.40 | AR | UCMD1 (LGMD22) 254090 | 2q37.3 | <i>COL6A3</i> 120250 | Collagen type VI subunit alpha 3 | Demir et al (2002) | allelic to LGMDD5 (group 1); UCMD1 and BTHLM1 (group 2) |
| Muscular dystrophy, limb-girdle, autosomal recessive 23 | 1.41 | AR | LGMDR23 618138 | 6q22.33 | <i>LAMA2</i> 156225 | Laminin 2, Heavy chain (laminin alpha2 chain of merosin) | Gavassini et al (2011) | allelic to MDC1A (group 2) |
| Muscular dystrophy, limb-girdle, autosomal recessive 25 | 1.42 | AR | LGMDR25 (LGMD2X) 616812 | 6q21 | <i>BVES</i> (= <i>POPDC1</i>) 604577 | Blood vessel epicardial substance | Schindler et al (2016) | |
| Muscular dystrophy, limb-girdle, autosomal recessive 26 | 1.43 | AR | LGMDR26 618848 | 6q21 | <i>POPDC3</i> 605824 | Popeye domain-containing protein 3 | Vissing et al (2019) | |
| Muscular dystrophy, limb-girdle, autosomal recessive 27 | 1.44 | AR | LGMDR27 619566 | 14q32.33 | <i>JAG2</i> 602570 | Jagged 2 | Coppens et al (2021) | |

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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) |
|---|-------------------------|-------------|---|--------------|-------------------------------------|--|---|--|
| Muscular dystrophy; limb-girdle, autosomal recessive 28 | 1.45 | AR | LGMDR28 620375 | 5q13.3 | HMGCR 142910 | 3-Hydroxy-3-methylglutaryl-CoA reductase | Yogev et al (2023) Morales-Rosado et al (2023) | |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C1 | 1.46 | AR | MDDGC1 (LGMDR11) 609308 | 9q34.13 | POMT1 607423 | Protein O-mannosyltransferase 1 | Balci et al (2005) D'Amico et al (2006) | Allelic to MDDGA1 (group 2) and MDDGB1 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C2 | 1.47 | AR | MDDGC2 (LGMDR14) 613158 | 14q24.3 | POMT2 607439 | Protein O-mannosyltransferase 2 | Biancheri et al (2007) | Allelic to MDDGA2 and MDDGB2(group 2) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C3 | 1.48 | AR | MDDGC3 (LGMDR15) 613157 | 1p34.1 | POMGNT1 606822 | Protein O-linked mannose beta1,2-N-acetylglucosaminyltransferase 1 | Godfrey et al (2007) Clement et al (2008) Raducu et al (2012) | Allelic to MDDGA3 and MDDGB3 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C4 | 1.49 | AR | MDDGC4 (LGMDR13) 611588 | 9q31.2 | FKTN 607440 | Fukutin | Murakami et al (2006) Godfrey et al (2006) | Allelic to MDDGA4 and MDDGB4 (group 2), CMD1X (group 10A) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C5 | 1.50 | AR | MDDGC5 (LGMDR9) 607155 | 19q13.32 | FKRP 606596 | Fukutin related protein | Driss et al (2000) Brockington et al (2001a) | Allelic to MDDGA5 and MDDGB5 (group 2), |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C7 | 1.51 | AR | MDDGC7 (LGMDR20) 616052 | 7p21.2-p21.1 | CRPPA 614631 | Isoprenoid synthase domain containing protein | Tasca et al (2013) | Allelic to MDDGA7 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C8 | 1.52 | AR | MDDGC8 (LGMDR24) 618135 | 3p22.1 | POMGNT2 614828 | Protein O-mannose beta-1,4-N-acetylglucosaminyltransferase 2 | Endo et al (2015) | allelic to MDDGA8 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C9 | 1.53 | AR | MDDGC9 (LGMDR16) 613818 | 3p21.31 | DAG1 128239 | Dystrophin-associated glycoprotein 1 (alpha-dystroglycan) | Hara et al (2011) | Allelic to MDDGA9 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C12 | 1.54 | AR | MDDGC12 616094 | 8p11.21 | POMK 615247 | Protein-O-mannose kinase | Di Costanzo et al. (2014) | Allelic to MDDGA12 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C14 | 1.55 | AR | MDDGC14 (LGMDR19) 615352 | 3p21.31 | GMPPB 615320 | GDP-mannose pyrophosphorylase B | Carss et al (2013) Cabrera-Serrano et al (2015) | Allelic to MDDGA14, MDDGB14 (group 2) and congenital myasthenic syndrome (group 11) |
| Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C15 | 1.56 | AR | MDDGC15 612937 | 1q22 | DPM3 605951 | Dolichyl-phosphate mannosyltransferase polypeptide 3 | Lefeber et al (2009) | Allelic to MDDGB15 (group 2) |
| Muscular dystrophy with cardiomyopathy and triangular tongue | 1.57 | AR | MDRCMTT (LGMD2W) 616827 | 2q14.3 | LIMS2 (=PINCH2) 607908 | LIM and senescent cell antigen-like domains 2 | Chardon et al (2015) | |
| Scapuloperoneal muscular dystrophy and dropped head syndrome | 1.58 | AR / AD | 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 | 1.59 | AR | | 8q24.3 | PLEC 601282 | Plectin | Fattahi et al (2015) | allelic to LGMDR17 (group 1), EBSMD (group 5), myasthenic syndrome related to <i>PLEC</i> (group 11) |
| LGMD related to <i>PYROXD1</i> | 1.60 | AR | | 12p12.1 | PYROXD1 617220 | Pyridine nucleotide-disulphide oxidoreductase domain 1 | Sainio et al (2018) | allelic to Congenital Myopathy related to <i>PYROXD1</i> (group 3), MMF8 (group 5) |
| LGMD related to <i>DTNA</i> | 1.61 | AD | | | DTNA 601239 | Dystrobrevin alpha | Nascimento et al (2023) | Allelic to LVNC1 (group 10A) |

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) 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) |

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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) |
|---|-------------------------|-------------|---|------------|--|--|---|--|
| Bethlem myopathy 1 | 2.4 | AD | BTHLM1 158810 | 2q37.3 | <i>COL6A3</i> 120250 | Collagen type VI subunit alpha 3 | Speer et al (1996) Bertini et al (1998) Pan et al (1998) Gualandi et al (2009) | allelic to LGMDD5 and LGMDR22 (group 1); UCMD1 (group 2) |
| Bethlem myopathy 1 | 2.5 | AR | BTHLM1 158810 | 21q22.3 | <i>COL6A2</i> 120240 | Collagen type VI subunit alpha 2 | | 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 | <i>COL6A1</i> 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 | <i>COL6A2</i> 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 | <i>COL6A3</i> 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 | <i>COL12A1</i> 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 | <i>COL12A1</i> 120320 | Collagen type XII alpha 1 chain | Zou et al (2014) | Allelic to UCMD2 (group 2), CMD related to COL12A1 (group 2) |
| <i>COL12A1</i> -related congenital muscular dystrophy | 2.11 | AD | | 6q13-q14 | <i>COL12A1</i> 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 | <i>COL6A2</i> 120240 | Collagen type VI subunit alpha 2 | Merlini et al (2008) | allelic to LGMDD5 and LGMDR22 (group 1); UCMD1 and BTHLM1 (group 2) |
| Congenital myopathy 3 with rigid spine | 2.13 | AR | CMYP3 602771 | 1p36.11 | <i>SELENON</i> (formerly <i>SEPN1</i>) 606210 | Selenoprotein N | Moghadaszadeh et al (1998, 2001) Ferreiro et al (2002, 2004) | |
| Congenital muscular dystrophy due to ITGA7 deficiency | 2.14 | AR | 613204 | 12q13.2 | <i>ITGA7</i> 600536 | Integrin alpha7 | Hayashi et al (1998) | |
| Congenital muscular dystrophy related to <i>DNM2</i> | 2.15 | AD | | 19p13.2 | <i>DNM2</i> 602378 | Dynamin 2 | Susman et al (2010) | allelic to CNM1 (group 3), Distal myopathy related to <i>DNM2</i> (group 4), LCCS5 (group 12) and CMTDIB (group 14) |
| Congenital muscular dystrophy related to <i>TCAP</i> | 2.16 | AR | | 17q12 | <i>TCAP</i> 604488 | Titin-cap (telethonin) | Ferreiro et al (2011) | allelic to LGMDR7 (group 1), CMH25 (group 10A), CMD1N (group 10A) |
| Congenital muscular dystrophy related to <i>LMNA</i> | 2.17 | AD | MDCL 613205 | 1q22 | <i>LMNA</i> 150330 | Lamin A/C | Quijano-Roy et al (2008) | Allelic to EDMD2, EDMD3 (group 1), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370] |
| <i>CMD-Dystroglycanopathies</i> | | | | | | | | |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A1 | 2.18 | AR | MDDGA1 236670 | 9q34.13 | <i>POMT1</i> 607423 | Protein-O-mannosyl transferase 1 | Beltran-Valero De Bernabe et al (2002) Mercuri et al (2009) | allelic to MDDGC1 (group 1), and MDDGB1 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A2 | 2.19 | AR | MDDGA2 613150 | 14q24.3 | <i>POMT2</i> 607439 | Protein O-mannosyl transferase 2 | van Reeuwijk et al (2005) Mercuri et al (2009) | Allelic to MDDGC2 (group 1) and MDDGA2, MDDGB2 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A3 | 2.20 | AR | MDDGA3 253280 | 1p34.1 | <i>POMGNT1</i> 606822 | Protein O-mannose beta1,2-N-acetylglucosaminyl transferase | Yoshida et al (2001) Taniguchi et al (2003) | Allelic to MDDGC3 (group 1) and MDDGB3 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A4 | 2.21 | AR | MDDGA4 253800 | 9q31.2 | <i>FKTN</i> 607440 | Fukutin | Toda et al (1993) Kobayashi et al (1998) | Allelic to MDDGC4 (group 1), MDDGB4 (group 2) and CMD1X (group 10A) |

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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) |
|---|-------------------------|-------------|---|------------|-----------------------------|---|---|--|
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A5 | 2.22 | AR | MDDGA5 613153 | 19q13.32 | <i>FKRP</i> 606596 | Fukutin related protein | Cormand et al (2001) Beltran-Valero De Bernabe et al (2004) | Allelic to MDDGC5 (group 1) and MDDGB5 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A6 | 2.23 | AR | MDDGA6 613154 | 22q12.3 | <i>LARGE</i> 603590 | Acetylglucosaminyl-transferase-like protein (like-glycosyl transferase) | Longman et al (2003) Mercuri et al (2009) | Allelic to MDDGB6 (group 2) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A7 | 2.24 | AR | MDDGA7 614643 | 7p21.2 | <i>CRPPA</i> 614631 | Isoprenoid synthase domain containing | Roscioli et al (2012) Willer et al (2012) | Allelic to MDDGC7 (group 1) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A8 | 2.25 | AR | MDDGA8 614830 | 3p22.1 | <i>POMGNT2</i> 614828 | Protein O-mannose beta-1,4-N-acetylglucosaminyl transferase 2 | Manzini et al (2012) | allelic to MDDGC8 (group 1) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A9 | 2.26 | AR | MDDGA9 616538 | 3p21.31 | <i>DAG1</i> 128239 | Dystrophin-Associated Glycoprotein 1 | Geis et al (2013) | allelic to MDDGC9 (group 1) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A10 | 2.27 | AR | MDGGA10 615041 | 12q14.2 | <i>RXYLT1</i> 605862 | Ribitol xylosyltransferase 1 (transmembrane protein 5) | Vuillaumier-Barrot et al (2013) | |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A11 | 2.28 | AR | MDDGA11 615181 | 1q42.3 | <i>B3GALNT2</i> 610194 | Beta-1,3-N-acetylgalactosaminyl-transferase 2 | Stevens et al (2013) | |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A12 | 2.29 | AR | MDDGA12 615249 | 8p11.21 | <i>POMK</i> 615247 | Protein-O-mannose kinase | Jae et al (2013) | Allelic to MDDGC12 (group 1) |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A13 | 2.30 | AR | MDDGA13 615287 | 11q13.2 | <i>B4GAT1</i> 605517 | Beta-1,4-glucuronyl transferase 1 (Beta-1,3-N-acetylglucosaminyl-transferase 1) | Buyse et al (2013) Shaheen et al (2013) | |
| Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A14 | 2.31 | AR | MDDGA14 615350 | 3p21.31 | <i>GMPPB</i> 615320 | GDP-mannose pyrophosphorylase B | de Carss et al (2013) | Allelic to MDDGC14 (group 1), MDDGB14 (group 2), congenital myasthenic syndrome (group 11) |
| Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B1 | 2.32 | AR | MDDGB1 613155 | 9q34.13 | <i>POMT1</i> 607423 | Protein-O-mannosyl transferase 1 | van Reeuwijk et al (2006) Mercuri et al (2009) | Allelic to MDDGC1 (group 1) and MDDGA1 (group 2) |
| Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B2 | 2.33 | AR | MDDGB2 613156 | 14q24.3 | <i>POMT2</i> 607439 | Protein O-mannosyl transferase 2 | Yanagisawa et al (2007) | Allelic to MDDGC2 (group 1) and MDDGA2 and MDDGB2 (group 2) |
| Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B3 | 2.34 | AR | MDDGB3 613151 | 1p34.1 | <i>POMGNT1</i> 606822 | Protein O-mannose beta1,2-N-acetylglucosaminyl transferase | Clement et al (2008) Mercuri et al (2009) | Allelic to MDDGC3 (group 1) and MDDGA3 (group 2) |
| Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B4 | 2.35 | AR | MDDGB4 613152 | 9q31.2 | <i>FKTN</i> 607440 | Fukutin | Beltran-Valero de Bernabe (2003) Mercuri et al (2009) | Allelic to MDDGC4 (group 1), MDDGA4 (group 2) and CMD1X (group 10A) |
| Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B5 | 2.36 | AR | MDDGB5 606612 | 19q13.32 | <i>FKRP</i> 606596 | Fukutin related protein | Brockington et al (2001b) Topaloglu et al (2003) Mercuri et al (2009) | Allelic to MDDGC5 (group 1) and MDDGA5 (group 2) |
| Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B6 | 2.37 | AR | MDDGB6 608840 | 22q12.3 | <i>LARGE</i> 603590 | Acetylglucosaminyl-transferase-like protein (like-glycosyl transferase) | Longman et al (2003) Mercuri et al (2009) | Allelic to MDDGA6 (group 2) |

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|---|-------------------------|-------------|---|--------------|---------------------------------|--|--|---|
| Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B14 | 2.38 | AR | MDDGB14 615351 | 3p21.31 | GMPPB 615320 | GDP-mannose pyrophosphorylase B | Carss et al (2013) | Allelic to MDDGC14 (group 1), MDDGA14 (group 2) and congenital myasthenic syndrome (group 11) |
| Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B15 | 2.39 | AR | MDDGB15 618992 | 1q22 | DPM3 605951 | | Fu et al (2019) | Allelic to MDDGC15 (group 1) |
| Muscle-eye-brain disease (MEB) | 2.40 | AD | MEB | 11q13.2q14.1 | 11q13.2q14.1 duplication | | Villar-Quiles et al (2020) | |
| Congenital disorder of glycosylation, type 1e | 2.41 | AR | CDG1E 608799 | 20q13.13 | DPM1 603503 | Dolichyl-phosphate mannosyltransferase 1, catalytic subunit | Kim et al (2000) Yang et al (2013) | |
| Congenital disorder of glycosylation, type 1f | 2.42 | AR | CDG1F 609180 | 17p13.1 | MPDU1 604041 | Mannose-P-dolichol utilization defect 1 | Schenk et al (2001) van Tol et al (2019) | |
| Congenital disorder of glycosylation, type 1u | 2.43 | AR | CDG1U 615042 | 9q34.11 | DPM2 603564 | Dolichyl-phosphate mannosyltransferase polypeptide 2, regulatory subunit | Barone et al (2012) | |
| Other congenital muscular dystrophies | | | | | | | | |
| Congenital muscle dystrophy with mitochondrial structural abnormalities (megaconial type) | 2.44 | AR | MDCMC 602541 | 22q13.33 | CHKB 612395 | Choline kinase beta | Mitsuhashi et al (2011) | |
| Congenital muscular dystrophy | 2.45 | AR | MDC1B 604801 | 1q42 | ? | | Brockington et al (2000) | |
| Developmental delay with hypotonia, myopathy and brain abnormalities | 2.46 | AR | DEDHMB 620240 | 9q34.11 | GOLGA2 602580 | Golgin A2 | Shamseldin et al (2016) | |
| Muscular dystrophy, congenital Davignon-Chauveau type | 2.47 | 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 | 2.48 | 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 <i>MSTO1</i> | 2.49 | AR | MMYAT617619 | 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) |
| Congenital muscular dystrophy related to <i>RYR1</i> | 2.50 | AR | | 19q13.2 | RYR1 180901 | Ryanodine receptor | Helbling et al (2019) | Allelic to CMYP1A, CMYP1B, CCD, CFTD, CNMDU1, Centronuclear myopathy related to RYR1, DuCD related to RYR1 (group 3), Late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia deformation sequence related to RYR1 (group 16) |
| Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by <i>TRAPPC11</i> mutations | 2.51 | 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 or without seizures | 2.52 | AR | MYOS 620166 | 17q21.32 | GOSR2 604027 | Golgi SNAP receptor complex member 2 | Tsai et al (2013) Larson et al (2018) | |
| Congenital muscular dystrophy with epilepsy | 2.53 | AR | | 7q21.3 | BET1 605456 | BET1 golgi vesicular membrane-trafficking protein | Donkervoort et al (2021) | |
| Congenital muscular dystrophy related to <i>TRAPPC2L</i> | 2.54 | AR | | 16q24.3 | TRAPPC2L 610970 | Trafficking protein particle complex 2-like | Abaji et al (2023) | |

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|---|-------------------------|-------------|---|------------|--|--|---|---|
| GROUP 3. CONGENITAL MYOPATHIES | | | | | | | | |
| <i>Nemaline myopathies</i> | | | | | | | | |
| Congenital myopathy 2A, typical | 3.1 | AD | CMYP2A (NEM3) 161800 | 1q42.13 | <i>ACTA1</i> 102610 | Actin, alpha 1, skeletal muscle | Agrawal et al (2004) | Allelic to CMYP2B, CMYP2C, CFTD and cap myopathy (group 3) |
| Congenital myopathy 2B, severe infantile | 3.2 | AR | CMYP2B 161800 | 1q42.13 | <i>ACTA1</i> 102610 | Actin, alpha 1, skeletal muscle | Nowak et al (1999) Sparrow et al (2003) | Allelic to CMYP2A, CMYP2C, CFTD and cap myopathy (group 3) |
| Congenital myopathy 2C, severe infantile, dominant | 3.3 | AD | CMYP2C 620278 | 1q42.13 | <i>ACTA1</i> 102610 | Actin, alpha 1, skeletal muscle | Goebel et al (1997) Nowak et al (1999) | Allelic to CMYP2A, CMYP2B, CMYP2C, CFTD and cap myopathy (group 3) |
| Congenital myopathy 4A, dominant | 3.4 | AD | CMYP4A (NEM1) 255310 | 1q21.3 | <i>TPM3</i> 191030 | Tropomyosin 3 | Laing et al. (1992) Laing et al. (1995b) Tan et al (1999) Wattanasirichaigoon et al (2002) | Allelic to CMYP4B (group 3) |
| Congenital myopathy 4B, recessive | 3.5 | AR | CMYP4B 609284 | 1q21.3 | <i>TPM3</i> 191030 | Tropomyosin 3 | Tan et al (1999) Wattanasirichaigoon et al (2002) | Allelic to CMYP4A (group 3) |
| Nemaline myopathy 2 | 3.6 | AR | NEM2 256030 | 2q23.3 | <i>NEB</i> 161650 | Nebulin | Wallgren-Pettersson et al (1995, 2002) Pelin et al (1999) Lehtokari et al (2006) | |
| Nemaline myopathy 4 | 3.7 | AD | NEM4 609285 | 9p13.3 | <i>TPM2</i> 190990 | Tropomyosin 2 (beta) | Donner et al (2002) Monnier et al (2009) | Allelic to CMYP23 (group 3), DA2B4 and DA1A (group 16) |
| Nemaline myopathy 5 | 3.8 | AR | NEM5 605355 | 19q13.42 | <i>TNNT1</i> 191041 | Troponin T type 1 (skeletal, slow) | Johnston et al (2000) | |
| Nemaline myopathy 6 | 3.9 | AD | NEM6 609273 | 15q22.31 | <i>KBTBD13</i> 613727 | Kelch repeat and BTB/POZ domain containing protein 13 | Gommans et al (2003) Samburghin et al (2010) | allelic to LGMD related to <i>KBTBD13</i> (group 1) |
| Nemaline myopathy 7 | 3.10 | AR | NEM7 610687 | 14q13.1 | <i>CFL2</i> 601443 | Cofilin 2 (muscle) | Agrawal et al (2007) | |
| Nemaline myopathy 8 | 3.11 | AR | NEM8 615348 | 3p22.1 | <i>KLHL40</i> 615340 | Kelch-like family member 40 | Ravenscroft et al (2013) | |
| Nemaline myopathy 9 | 3.12 | AR | NEM9 615731 | 2q31.1 | <i>KLHL41</i> 607701 | Kelch-like family member 41 | Gupta et al (2013) | |
| Nemaline myopathy 10 | 3.13 | AR | NEM 10 616165 | 3p14.1 | <i>LMOD3</i> 616112 | Leiomodin 3 | Yuen et al (2014) | |
| Nemaline myopathy 11 | 3.14 | AR | NEM11 617336 | 10q21.3 | <i>MYPN</i> 608517 | Myopalladin | Miyatake et al (2016) Lornage et al (2017) | allelic to CMD1KK, CMH22 and RCM4 (group 10A) |
| Klippel-Feil syndrome with Nemaline myopathy and facial dysmorphism | 3.15 | AR | KFS4 616549 | 22q12.1 | <i>MYO18B</i> 607295 | Myosin XVIIIIB | Malfatti et al (2015) | |
| Congenital myopathy 20 | 3.16 | AR | CMYP20 620310 | 15q13-q14 | <i>RYR3</i> 180903 | Ryanodine receptor 3 | Nilipour et al (2018) | |
| Nemaline myopathy | 3.17 | AR | | 11p15.5 | <i>TNNT3</i> 600692 | Troponin T3, fast skeletal | Sandaradura et al (2018) | allelic to DA2B2 (group 16) |
| <i>Other congenital myopathies</i> | | | | | | | | |
| Myopathy, centronuclear, X-linked | 3.18 | XLR | CNMX 310400 | Xq28 | <i>MTM1</i> 300415 | Myotubularin 1 | Thomas et al (1987) Laporte (1996, 1997, 2000) | |
| Myopathy centronuclear 1 | 3.19 | AD | CNM1 160150 | 19p13.2 | <i>DNM2</i> 602378 | Dynamin 2 | Bitoun et al (2005) | allelic to CMD related to <i>DNM2</i> (group 2), CNM1 (group 4), LCCS5 (group 12) and CMTDIB (group 14) |
| Myopathy centronuclear 2 | 3.20 | AR, AD | CNM2 255200 | 2q14.3 | <i>BIN1</i> 601248 | Bridging integrator 1 (Amphiphysin) | Nicot et al (2007) Böhm et al (2014) | |
| Myopathy centronuclear 4 | 3.21 | AD | CNM4 614807 | 16p13.3 | <i>CCDC78</i> 614666 | Coiled-coil domain-containing protein 78 | Majczenko et al (2012) | |
| Myopathy centronuclear 5 | 3.22 | AR | CNM5 615959 | 2q35 | <i>SPEG</i> 615950 | SPEG complex locus | Agrawal et al (2014) | |
| Myopathy centronuclear 6 with fiber-type disproportion | 3.23 | AR | CNM6 617760 | 3p23-21 | <i>MAP3K20</i> (formerly <i>ZAK</i>) 609479 | Mitogen-activated protein 3 kinase 20 | Tetreault et al (2006) Vasli et al (2017) | |

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|---|-------------------------|-------------|---|------------|-----------------------------|--|---|--|
| Centronuclear myopathy related to <i>RYR1</i> | 3.24 | AR | | 19q13.2 | <i>RYR1</i> 180901 | Ryanodine receptor | Wilmshurst et al (2010) | allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYP1A</i> , <i>CMYP1B</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16) |
| Centronuclear myopathy (CNM) related to <i>TTN</i> | 3.25 | AR | | 2q31.2 | <i>TTN</i> 188840 | Titin | Ceyhan-Birsoy et al (2013) | allelic to <i>LGMDR10</i> (group 1), <i>MmD</i> related to <i>TTN</i> and <i>CMYP5</i> (group 3), <i>TMD</i> (group 4), <i>MFM9</i> (group 5), <i>CMH9</i> (group 10), <i>CMD1G</i> (group 10), <i>LCCS</i> related to <i>TTN</i> (group 12) |
| Congenital myopathy 1A, dominant, with susceptibility to malignant hyperthermia | 3.26 | AD | <i>CMYP1A</i> 117000 | 19q13.2 | <i>RYR1</i> 180901 | Ryanodine receptor | Kausch et al (1991) Zhang et al (1993) Quane et al. (1993) Robinson et al (2002) | allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYP1B</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16) |
| Congenital myopathy 1B, recessive Minicore myopathy | 3.27 | AR | <i>CMYP1B</i> 255320 | 19q13.2 | <i>RYR1</i> 180901 | Ryanodine receptor | Monnier et al (2003) Jungbluth et al (2005) | allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYP1A</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16) |
| Central core disease, recessive (transient multimimicore myopathy) | 3.28 | AR | <i>CCD</i> 117000 | 19q13.2 | <i>RYR1</i> 180901 | Ryanodine receptor | Ferreiro et al (2002a) Jungbluth et al (2002) | allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYP1A</i> , <i>CMYP1B</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16) |
| Multimimicore disease (MmD) related to <i>TTN</i> | 3.29 | AR | | 2q31.2 | <i>TTN</i> 188840 | Titin | Ge et al (2019) | allelic to <i>LGMDR10</i> (group 1), <i>CNM</i> related to <i>TTN</i> and <i>CMYP5</i> (group 3), <i>TMD</i> (group 4), <i>MFM9</i> (group 5), <i>CMH9</i> (group 10A), <i>CMD1G</i> (group 10A), <i>LCCS</i> related to <i>TTN</i> (group 12) |
| Congenital myopathy 5 with cardiomyopathy (formerly Salih myopathy) | 3.30 | AR | <i>CMYP5</i> (<i>SALMY</i>) 611705 | 2q31.2 | <i>TTN</i> 188840 | Titin | Carmignac et al (2007) | allelic to <i>LGMDR10</i> (group 1), <i>CNM</i> related to <i>TTN</i> and <i>MmD</i> related to <i>TTN</i> (group 3), <i>TMD</i> (group 4), <i>MFM9</i> (group 5), <i>CMH9</i> (group 10), <i>CMD1G</i> (group 10), <i>LCCS</i> related to <i>TTN</i> (group 12) |
| Congenital myopathy 6 with ophtalmoplegia | 3.31 | AD, AR | <i>CMYP6</i> 605637 | 17p13.1 | <i>MYH2</i> 160740 | Myosin, heavy chain 2, skeletal muscle, adult | Martinsson et al (1999, 2000) Tajsharghi et al (2010) | |
| Congenital myopathy 7A, myosin storage, autosomal dominant | 3.32 | AD | <i>CMYP7A</i> 608358 | 14q11.2 | <i>MYH7</i> 160760 | Myosin, heavy chain 7, cardiac muscle, beta | Tajsharghi et al (2003) Bohlega et al (2004) Laing et al (2005) | allelic to <i>CFTD</i> , <i>CMYP7B</i> , <i>MSM</i> with eccentric core disease (group 3), <i>MPD1</i> (group 4), <i>CMH1</i> , <i>CMD1S</i> and <i>LVNC5</i> (group 10A) |
| Congenital myopathy 7B, myosin storage, autosomal recessive | 3.33 | AR | <i>CMYP7B</i> 255160 | 14q11.2 | <i>MYH7</i> 160780 | Myosin, heavy chain 7, cardiac muscle, beta | Onengut et al (2004) Tajsharghi et al (2007) Yuceyar et al (2015) | allelic to <i>CFTD</i> , <i>CMYP7A</i> , <i>MSM</i> with eccentric core disease and <i>SPMM</i> related to <i>MYH7</i> (group 3), <i>MPD1</i> (group 4), <i>CMH1</i> , <i>CMD1S</i> and <i>LVNC5</i> (group 10A) |

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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) |
|--|-------------------------|-------------|---|------------|---|---|--|--|
| Congenital myopathy 8 | 3.34 | AD | CMYP8 618654 | 1q43 | <i>ACTN2</i> 102573 | Actinin, alpha 2 | Lornage et al (2019) | allelic to MPD6 (group 4), CMH23 and CMD1AA (group 10A) |
| Congenital myopathy 9A | 3.35 | AR | CMYP9A 618822 | 3q26.33 | <i>FXR1</i> <i>600819</i> | FMR1 autosomal homolog | Estan et al (2019) | allelic to CMYP9B (group 3) |
| Congenital myopathy 9B, proximal with minicore | 3.36 | AR | CMYP9B 618823 | 3q26.33 | <i>FXR1</i> <i>600819</i> | FMR1 autosomal homolog | Estan et al (2019) | allelic to CMYP9A (group 3) |
| Congenital myopathy 10A, severe variant | 3.37 | AR | CMYP10A 614399 | 5q23.2 | <i>MEGF10</i> <i>612453</i> | Multiple EGF-like-domains 10 | Logan et al (2011) | allelic to CMYP10B (group 3) |
| Congenital myopathy 10B, mild variant | 3.38 | AR | CMYP10B 620249 | 5q23.2 | <i>MEGF10</i> <i>612453</i> | Multiple EGF-like-domains 10 | Boyden et al (2012) | allelic to CMYP10A (group 3) |
| Congenital Myopathy 11 | 3.39 | AR | CMYP11 619967 | 10p12.33 | <i>HACD1</i> (<i>PTPLA</i>) <i>610467</i> | 3-Hydroxyacyl-CoA dehydratase (Protein tyrosine phosphatase-like) | Muhammad et al (2013) | allelic to ARVD6 (group 10B) |
| Congenital myopathy 12 | 3.40 | AR | CMYP12 612540 | 12q12 | <i>CNTN1</i> <i>600016</i> | Contactin-1 | Compton et al (2008) | |
| Congenital myopathy 13 | 3.41 | AR | CMYP13 255995 | 12q13.3 | <i>STAC3</i> <i>615521</i> | SH3 and cysteine-rich domains 3 | Horstick et al (2013) | allelic to Myopathy, congenital, with malignant hyperthermia susceptibility (group 3) |
| Congenital myopathy 14 | 3.42 | AR | CMYP14 618414 | 2q34 | <i>MYL1</i> <i>160780</i> | Myosin, light polypeptide 1, alkali, skeletal fast | Ravenscroft et al (2018) | |
| Congenital myopathy 15 | 3.43 | AD | CMYP15 620161 | 20q13.12 | <i>TNNC2</i> <i>191039</i> | Troponin C Fast | van de Locht (2021) | |
| Congenital myopathy 16 | 3.44 | AD | CMYP16 618524 | 12q23.2 | <i>MYBPCI</i> <i>160794</i> | Myosin-binding protein C, slow type | Shashi et al (2019) | allelic to LCCS4 (group 12), DA1B (group 16) |
| Congenital myopathy 17 | 3.45 | AR | CMYP17 618975 | 11p15.1 | <i>MYOD1</i> <i>159970</i> | Myogenic Differentiation Antigen 1 | Watson et al (2016) | |
| Congenital myopathy 18 due to dihydropyridine receptor defect | 3.46 | AD, AR | CMYP18 620246 | 1q32.1 | <i>CACNA1S</i> <i>114208</i> | Calcium channel, voltage-dependent, L type, alpha 1S subunit | Schartner et al (2019) | allelic to LGMD related to CACNA1S (group 1), HOKPP1 (group 7) and MHS5 (group 8) |
| Congenital myopathy 19 | 3.47 | AR | CMYP19 618578 | 1p36.13 | <i>PAX7</i> <i>167410</i> | Paired Box gene 7 | Feichtinger et al (2019) | |
| Congenital Myopathy 22A, classic | 3.48 | AR | CMYP22A 620351 | 17q23.3 | <i>SCN4A</i> <i>603967</i> | Sodium channel, voltage-gated, type IV, alpha subunit | Zaharieva et al (2016) Gonorazky et al (2017) Sloth et al (2018) | allelic to CMYP22B (group 3), HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16) |
| Congenital Myopathy 22B, severe fetal | 3.49 | AR | CMYP22B 620369 | 17q23.3 | <i>SCN4A</i> <i>603967</i> | Sodium channel, voltage-gated, type IV, alpha subunit | Zaharieva et al (2016) | allelic to CMYP22A (group 3), HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16) |
| Congenital myopathy 23 (Cap myopathy) | 3.50 | AD | CMYP23 609285 | 9p13.3 | <i>TPM2</i> <i>190990</i> | Tropomyosin 2, beta | Tajsharghi et al (2007) Lehtokari et al (2007) | allelic to NEM4 (group 3), DA1A and DA2B4 (group 16) |
| Myopathy, myosin storage, autosomal dominant (eccentric core disease) | 3.51 | AD | | 14q11.2 | <i>MYH7</i> <i>160760</i> | Myosin, heavy chain 7, cardiac muscle, beta | Romero et al (2014) | allelic to CFTD, CMYP7A, CMYP7B (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10A) |
| Cap myopathy | 3.52 | AD | | 1q42.13 | <i>ACTA1</i> <i>102610</i> | Actin, alpha 1, skeletal muscle | Hung et al (2010) | allelic to CMD with rigid spine (group 2), CMYP2A, CMYP2B, CMYP2C and CFTD (group 3) |
| Congenital skeletal myopathy and fatal cardiomyopathy | 3.53 | AR | | 11p11.2 | <i>MYBPC3</i> <i>600958</i> | Cardiac myosin binding protein-C | Tajsharghi et al (2010) | allelic to CMH4, CMD1MM and LVNC10 (group 10A) |
| Congenital myopathy with ophthalmoplegia related to CACNA1S | 3.54 | AR | | 1q32.1 | <i>CACNA1S</i> <i>114208</i> | Calcium channel, voltage-dependent, L type, alpha 1S subunit | Hunter et al. (2015) | allelic to LGMD related to CACNA1S (group 1), CMYP18 (group 3), HOKPP1 (group 7) and MHS5 (group 8) |
| Carey-Fineman-Ziter syndrome (formerly congenital myopathy with Moebius sequence and Robin sequence) | 3.55 | AR | CFZS 254940 | 9q34.2 | <i>MYMK</i> <i>615345</i> | Myomaker | Di Gioia et al (2017) | |
| Carey-Fineman-Ziter syndrome 2 | 3.56 | AR | CFZS2 619941 | 6p21.1 | <i>MYMX</i> <i>619912</i> | Myomixer | Ramirez-Martinez et al. (2022) | |

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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) |
|---|-------------------------|-------------|---|-----------------------|-----------------------------|---|--|--|
| Myopathy, congenital, with malignant hyperthermia susceptibility | 3.57 | AR | | 12q13.3 | <i>STAC3</i> 615521 | SH3 and cysteine-rich domains 3 | Zaharieva et al (2018) | allelic to CMYP13 (group 3) |
| Congenital Myopathy related to <i>PYROXD1</i> | 3.59 | AR | | 12p12.1 | <i>PYROXD1</i> 617220 | Pyridine nucleotide-disulphide oxidoreductase domain 1 | Lornage et al (2019) | allelic to LGMD related to <i>PYROXD1</i> (group 1) and MMF8 (group 5) |
| Dusty core disease related to RYR1 | 3.60 | AR | DuCD | 19q13.2 | <i>RYR1</i> 180901 | Ryanodine receptor I | Garibaldi et al (2019) | allelic to Congenital muscular dystrophy related to RYR1 (group 2), CMYP1A, CMYP1B, CCD, CFTD, CNMDU1, Centronuclear myopathy related to RYR1 (group 3), Late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia deformation sequence related to RYR1 (group 16) |
| GROUP 4. DISTAL MYOPATHIES | | | | | | | | |
| Miyoshi muscular dystrophy 1 | 4.1 | AR | MMD1 254130 | 2p13.2 | <i>DYSF</i> 603009 | Dysferlin | Bejaoui et al . (1995) Bashir et al (1998) Liu et al (1998) | allelic to LGMDR2 (group 1) |
| Miyoshi muscular dystrophy 2 | 4.2 | AR | MMD2 613318 | 8q22.3 | | | Linssen et al (1998) | |
| Miyoshi muscular dystrophy 3 | 4.3 | AR | MMD3 613319 | 11p14.3 | <i>ANOS</i> 608662 | Anoctamin 5 | Bolduc et al. (2010) | allelic to LGMDR12 (group 1) and Muscular dystrophy with gnathodiaphyseal dysplasia (group 1) |
| Tibial muscular dystrophy (Udd) | 4.4 | AD | TMD 600334 | 2q31.2 | <i>TTN</i> 188840 | Titin | Haravuori et al (1998, 2001) Hackman et al (2002) | allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> , MmD related to <i>TTN</i> and CMYP5 (group 3), MFM9 (group 5), CMH9 (group 10), CMD1G (group 10), LCCS related to <i>TTN</i> (group 12) |
| Nonaka Myopathy | 4.5 | AR | NM 605820 | 9p13.3 | <i>GNE</i> 603824 | Glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase | Mitrani-Rosenbaum et al (1996) Ikeuchi et al (1997) Eisenberg et al (2001) | |
| Myopathy, Distal, 1 (Laing) | 4.6 | AD | MPD1 160500 | 14q11.2 | <i>MYH7</i> 160760 | Myosin, heavy chain 7, cardiac muscle, beta | Laing et al. (1995) Mastaglia et al (2000) Meredith et al (2004) | allelic to CFTD, CMYP7A, CMYP7B, MSM with eccentric core disease (group 3), CMH1, CMD1S and LVNC5 (group 10A) |
| Myopathy distal 3 | 4.7 | AD | MPD3 610099 | 8p22-q11 12q13-q22 | ? | ? | Haravuori et al (2004) | |
| Myopathy, distal, 4 | 4.8 | AD | MPD4 614065 | 7q32.1 | <i>FLNC</i> 102565 | Filamin C (gamma) | Duff et al (2011) | allelic to MFM5 (group 5), CMH26 and RCM5 (group 10A) |
| Myopathy, distal, 5 | 4.9 | AR | MPD5 617030 | 14q32.33 | <i>ADSSLI</i> 612498 | Adenylosuccinate synthase-like | Park et al (2016) | |
| Myopathy, distal 6, Adult-onset | 4.10 | AD | MPD6 618655 | 1q43 | <i>ACTN2</i> 102573 | Actinin alpha-2 | Savarese et al (2019) | allelic to CMYP8 (group 3), CMH23 and CMD1AA (group 10A) |
| Myopathy, distal, 7 adult onset, X-linked | 4.11 | XLD | MPD7 301075 | Xp22.12 | <i>SMPX</i> 300226 | Small Muscle Protein, X-linked | Johari et al (2021) | |
| Myopathy distal, Tateyama type | 4.12 | AD | MPDT 614321 | 3p25.3 | <i>CAV3</i> 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) |
| Vocal cord and pharyngeal distal myopathy (VCPDM) reclassified as ALS21 | 4.13 | AD | ALS21 606070 | 5q31.2 | <i>MATR3</i> 164015 | Matrin 3 | Feit et al (1998) Senderek et al (2009) | Allelic to ALS21 (group 12) |
| Welander distal myopathy | 4.14 | AD | WDM 604454 | 2p13.3 | <i>TIAI</i> 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) |

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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) |
|--|-------------------------|-------------|---|------------------|--|--|--|--|
| Welander-like distal myopathy | 4.15 | Digenic | | 5q35.3 2p13.3 | <i>SQSTM1</i> 601530 + | Sequestosome 1 + | Lee et al (2018) | <i>SQSTM1</i> Allelic to DMRV (group 4) and FTDALS3 (group 12) |
| Distal myopathy with myotilin defect | 4.17 | AD | | 5q31.2 | <i>TIA1</i> 603518 <i>MYOT</i> 604103 | Cytotoxic granule-associated RNA binding protein Myotilin | Penisson-Besnier et al (1998, 2006) | <i>TIA1</i> Allelic to WDM (group 4) allelic to MFM3 (group 5), spheroid body myopathy (group 5) |
| Distal myopathy with nebulin defect | 4.18 | AR, AD | | 2q23.3 | <i>NEB</i> 161650 | Nebulin | Wallgren-Pettersson et al (2007) Kiiiski et al (2019) | allelic to NEM2 (group 3) |
| Late onset distal myopathy (Markesbery-Griggs) | 4.19 | AD | | 10q23.2 | <i>LDB3</i> 605906 | LIM domain binding-3 | Griggs et al (2007) | allelic to MFM4 (group 5), CMD1C, CMH24 and LVNC3 (group 10A) |
| Distal myopathy related to <i>DNM2</i> | 4.20 | AD | | 19p13.2 | <i>DNM2</i> 602378 | Dynamamin 2 | Fischer et al (2006) | allelic to CMD related to <i>DNM2</i> (group 2), CNM1 (group 3), LCCS5 (group 12) and CMTDIB (group 14) |
| Early onset distal myopathy with <i>KLHL9</i> defect | 4.21 | AD | | 9p21.3 | <i>KLHL9</i> 611201 | Kelch-like 9 | Cirak et al (2010) | |
| Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 1 | 4.22 | AD | IBMPFD1 167320 | 9p13.3 | <i>VCP</i> 601023 | Valosin-containing protein | Palmio et al (2011) | allelic to Scapulo-peroneal myopathy (group 1), IBMPFD1 (group 5), ALS14 (group 12) and CMT2Y (group 14) |
| Myopathy, distal, with rimmed vacuoles | 4.23 | AD | DMRV 617158 | 5q35.3 | <i>SQSTM1</i> 601530 | Sequestosome 1 | Bucelli et al (2015) | allelic to Welander-like syndrome (group 4) and FTDALS3 (group 12) |
| Myopathy, distal, with rimmed vacuoles | 4.24 | AD | | 7q36.3 | <i>DNAJB6</i> 611332 | Hsp40 homologue, subfamily B, number 6 | Ruggieri et al (2015) | allelic to LGMDD1 (group 1) and distal myopathy (group 4) |
| Distal myopathy | 4.25 | AD | | 7q36.3 | <i>DNAJB6</i> 611332 | Hsp40 homologue, subfamily B, number 6 | Palmio et al (2019) | Allelic to LGMDD1 (group 1), Rimmed vacuole distal myopathy (group 4) |
| Distal Myopathy | 4.26 | AD | | 19p13.3 | <i>PLIN4</i> 613247 | Perilipin 4 | Ruggieri et al (2020) | |
| Rimmed vacuole myopathy | 4.27 | AD | | 12q24.23 | <i>HSPB8</i> 608014 | Heat-shock 22kD protein 8 | Al-Tahan et al (2019) | allelic to distal myopathy and motor neuropathy (group 4) HMND2 (group 12) and CMT2L (group 14) |
| Distal Myopathy and motor neuropathy | 4.28 | AD | | 12q24.23 | <i>HSPB8</i> 608014 | Heat-shock 22kD protein 8 | Ghaoui et al (2016) | Allelic to Rimmed vacuole myopathy (group 4), HMN2A (group 12) and CMT2L (group 14) |

GROUP 5. OTHER MYOPATHIES

| <i>Myofibrillar myopathies</i> | | | | | | | | |
|--------------------------------|-----|--------|----------------|----------|------------------------|------------------------------|--|---|
| Myofibrillar myopathy 1 | 5.1 | AD, AR | MFM1 601419 | 2q35 | <i>DES</i> 125660 | Desmin | Goldfarb et al (1998) Munoz-Marmol et al (1998) | Allelic to MFM1 with ARCV7 (group 5), CMD11 (group 10A), ARVC7 (group 10B), myopathy, cardiomyopathy and CMS (group 11) |
| Myofibrillar myopathy 2 | 5.2 | AD | MFM2 608810 | 11q23.1 | <i>CRYAB</i> 123590 | Alpha-B crystallin | Vicart et al (1998) Selcen et al (2003) | Allelic to CMD11I (group 10A) and CMT2 related to <i>CRYAB</i> (group 14) |
| Myofibrillar myopathy 3 | 5.3 | AD | MFM3 609200 | 5q31.2 | <i>MYOT</i> 604103 | Myotilin | Selcen and Engel (2004) | Allelic to distal myotilinopathy (group 4), spheroid body myopathy (group 5) |
| Spheroid body myopathy | 5.4 | AD | 182920 | 5q31.2 | <i>MYOT</i> 604103 | Myotilin | Foroud et al (2005) | allelic to distal myotilinopathy (group 4), MFM3 (group 5) |
| Myofibrillar myopathy 4 | 5.5 | AD | MFM4 609452 | 10q23.2 | <i>LDB3</i> 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 | <i>FLNC</i> 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 | <i>BAG3</i> 603883 | BCL2-associated athanogene 3 | Selcen et al (2009) | allelic to CMD1IH (group 10A) and CMT related to BAG3 (group 14) |

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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) |
|---|-------------------------|-------------|---|--------------------|--|---|---|--|
| Myofibrillar myopathy 7 | 5.8 | AR | MFM7 617114 | 3q22.2 | <i>KY</i> 605739 | Kyphoscoliosis peptidase | Hedberg-Oldfors et al (2016) Straussberg et al (2016) | |
| Myofibrillar myopathy 8 | 5.9 | AR | MFM8 617258 | 12p12.1 | <i>PYROXD1</i> 617220 | Pyridine nucleotide-disulphide oxidoreductase domain 1 | O'Grady et al (2016) | allelic to LGMD related to <i>PYROXD1</i> (group 1) and Congenital Myopathy related to <i>PYROXD1</i> (group 3) |
| Myofibrillar myopathy 9 with early respiratory failure | 5.10 | AD | MFM9 (HMERF) 603689 | 2q31.2 | <i>TTN</i> 188840 | Titin | Nicolao et al (1999) Lange et al (2005) | allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> , MmD related to <i>TTN</i> and CMYP5 (group 3), TMD (group 4), CMH9 (group 10A), CMD1G (group 10A), LCCS related to <i>TTN</i> (group 12) |
| Myofibrillar myopathy 10 | 5.11 | AR | MFM10 619040 | 10p11.23 | <i>SVIL</i> 604126 | Supervillin | Hedberg-Oldfors et al (2020) | |
| Myofibrillar myopathy 11 | 5.12 | AR | MFM11 619178 | 17q12 | <i>UNC45B</i> 611220 | UNC45 Myosin Chaperone B | Dafsari et al (2019) | |
| Myopathy, myofibrillar 12, infantile onset, with cardiomyopathy | 5.13 | AR | MFM12 619424 | 12q24.11 | <i>MYL2</i> 160781 | Myosin regulatory light chain 2 | Weterman et al (2013) | allelic to CMH10 (group 10A) |
| Cardiac and skeletal aggregate myopathy | 5.14 | Digenic | | 1p36.11 2p.23.3 | <i>TRIM63</i> 06131 + <i>TRIM54</i> 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 | 5.15 | AD | MFM1 601419 | 10q22 > 2q35 | <i>DES</i> 125660 | Desmin | Melberg et al (1999) Kuhl et al (2008) Hedberg et al (2012) | Allelic to MFM1 (group 5), CMDII (group 10A), ARVC7 (group 10B) and myopathy, cardiomyopathy and CMS (group 11) |
| Congenital myopathy 21 with early respiratory failure | 5.16 | AR | CMYP21 620326 | 1p31.1 | <i>DNAJB4</i> 611327 | DNAJ/HSP40 homolog, subfamily B, member 4 | Wheil et al (2022) | |
| Myofibrillar myopathy related to <i>FILIP1</i> | 5.17 | AR | | 6q14.1 | <i>FILIP1</i> 607307 | Filamin A-interacting protein 1 | Roos et al (2023) | |
| Miscellaneous | | | | | | | | |
| Danon disease | 5.18 | XLD | GSD2B 300257 | Xq24 | <i>LAMP2</i> 309060 | Lysosomal-associated membrane protein 2 | Nishino et al (2000) Musumeci et al (2005) | |
| Myopathy with excessive autophagy | 5.19 | XLR | MEAX 310440 | Xq28 | <i>VMA21</i> 300913 | <i>S. Cerevisiae</i> 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) Cortese et al (2014) | |
| Autophagic vacuolar myopathy | 5.20 | AR | CLN3 204200 | 16p12.1 | <i>CLN3</i> 607042 | Ceroid-lipofuscinosis, neuronal 3 (= Battenin) | | |
| Oculopharyngeal muscular dystrophy | 5.21 | AD | OPMD 164300 | 14q11.2 | <i>PABPN1</i> 602279 | Polyadenylate-binding protein, nuclear 1 | Brais et al (1995, 1998) Robinson et al (2005) | |
| Oculopharyngeal muscular dystrophy 2 | 5.22 | AD | OPMD2 620460 | 7p15.2 | <i>HNRNPA2B1</i> 600124 | Heterogeneous nuclear ribonucleoprotein A2/B1 | Kim et al (2022) | Allelic to IBMPFD2 (group 5) |
| Oculopharyngodistal myopathy 1 | 5.23 | AD | OPDM1 164310 | 8q22.3 | <i>LRP12</i> 618299 | Low density lipoprotein receptor-related protein 12 | Ishiura et al (2019) | Allelic to ALS28 (group 12) |
| Oculopharyngodistal myopathy 2 | 5.24 | AD | OPDM2 618940 | 19p13.12 | <i>GIPC1</i> 605072 | GIPC PDZ Domain-containing family, member 1 | Deng et al (2020) | |
| Oculopharyngodistal myopathy 3 | 5.25 | AD | OPDM3 619473 | 1q21.2 | <i>NOTCH2NLC</i> (<i>NBPF19</i>) 618025 | N-terminal-like protein | Ogasawara et al (2020) Yu et al (2021) | Allelic to Neuronal intranuclear inclusion diseases (group 14) and CMT2 related to NOTCH2NLC (group 14) |
| Oculopharyngodistal myopathy 4 | 5.26 | AD | OPDM4 619790 | 12q24.31 | <i>RILPL1</i> 614092 | Rab-interacting lysosomal protein-like 1 | Yu et al (2022) Zeng et al (2022) | |

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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) |
|--|-------------------------|-------------|---|------------|-----------------------------|--|---|--|
| Epidermolysis bullosa simplex associated with muscular dystrophy | 5.27 | AR | EBSMD 226670 | 8q24.3 | <i>PLEC</i> 601282 | Plectin | Gache et al (1996) Smith et al (1996) Wuyts et al (1996) | allelic to LGMDR17 (group 1), LGMD with ophtalmoplegia (group 1), myasthenic syndrome related to <i>PLEC</i> (group 11) |
| Muscle hypertrophy | 5.28 | AR | MSLHP 614160 | 2q32.2 | <i>MSTN</i> 601788 | Myostatin | Schuelke et al (2004) | |
| Fibrodysplasia ossificans progressiva | 5.29 | AD | FOP 135100 | 2q24.1 | <i>ACVRI</i> 102576 | Activin A receptor, type 1 | Shore et al (2006) | |
| Creatine phosphokinase, elevated serum (formerly HyperCKemia, idiopathic) | 5.30 | AD | 123320 | 3p25.3 | <i>CAV3</i> 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 | 5.31 | XLR | XMPMA 300696 | Xq26.3 | <i>FHLI</i> 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 | 5.32 | XLD | SPM 300695 | Xq26.3 | <i>FHLI</i> 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) | 5.33 | XLD | RBMX1A 300717 RBMX1B 300718 | Xq26.3 | <i>FHLI</i> 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) |
| Episodic muscle weakness, X-linked | 5.34 | XLR | EMWX 300211 | Xp22.3 | ? | ? | Ryan et al (1999) | |
| Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia 1 | 5.35 | AD | IBMPFD1 167320 | 9p13.3 | <i>VCP</i> 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 | 5.36 | AD | IBMPFD2 615422 | 7p15.2 | <i>HNRNPA2B1</i> 600124 | Heterogeneous nuclear ribonucleoprotein A2/B1 | Kim et al (2013) | Allelic to OPMD related to <i>HNRNPA2B1</i> (group 5) |
| Isolated inclusion body myopathy | 5.37 | AD | IBMPFD3 615424 | 12q13.13 | <i>HNRNPA1</i> 164017 | Heterogeneous nuclear ribonucleoprotein A1 | Izumi et al (2015) | allelic to ALS20 (group 12) |
| Myopathy with lactic acidosis, hereditary | 5.38 | AR | HML 255125 | 12q23.3 | <i>ISCU</i> 611911 | Iron-sulfur cluster scaffold homolog (E. coli) | Mochel et al (2008) | |
| Late onset axial myopathy related to <i>RYR1</i> | 5.39 | AD | | 19q13.2 | <i>RYR1</i> 180901 | Ryanodine receptor 1 (skeletal) | Løseth et al (2013) | Allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYP1A</i> , <i>CMYP1B</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , DuCD related to <i>RYR1</i> (group 3), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16) |
| Tubular aggregate myopathy 1 | 5.40 | AD | TAM1 160565 | 11p15.4 | <i>STIM1</i> 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 | 5.41 | AD | TAM2 615883 | 12q24.31 | <i>ORAI1</i> 610277 | ORAI calcium release-activated calcium modulator 1 | Nesin et al (2014) | |
| Sarcotubular myopathy | 5.42 | AR | | 9q33.1 | <i>TRIM32</i> 602290 | Tripartite motif containing 32 | Schoser et al (2005) | allelic to LGMDR8 (group 1) |
| Myopathy, sarcoplasmic body | 5.43 | AD | MYOSB 620286 | 22q12.3 | <i>MB</i> 160000 | Myoglobin | Olivé et al (2019) | |
| Vacuolar myopathy with CASQ1 aggregates | 5.44 | AD | VMCQA 616231 | 1q23.2 | <i>CASQ1</i> 114250 | Calsequestrin 1 | Rossi et al (2014) | |
| Lysosomal storage myopathy | 5.45 | AR | ML4 252650 | 19p13.2 | <i>MCOLN1</i> 605248 | Mucopilin 1 | Zambon et al. (2021) | |
| Myopathy, congenital, with excess of muscle spindles | 5.46 | AD | CSTLO 218040 | 11p15.5 | <i>HRAS</i> 190020 | V-Ha-RAS Harvey rat sarcoma viral oncogene homolog | Quélin et al (2017) | |
| Myopathy with extrapyramidal signs | 5.47 | AR | MPXPS 615673 | 10q22.1 | <i>MICU1</i> 605084 | Mitochondrial Calcium Uptake Protein 1 | Logan et al (2014) | |
| Muscle weakness with hyperlaxity, orofacial abnormalities and respiratory infections | 5.48 | AR | | 11p15.4 | <i>STIM1</i> 605921 | Stromal interaction molecule 1 | Salvi et al (2021) | Allelic to TAM1 (group 5) |

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|--|-------------------------|-------------|---|------------|-----------------------------|--|--|--|
| Myopathy with myalgia, increased serum creatine kinase and with or without episodic rhabdomyolysis | 5.49 | AR | MMCKR 620138 | 6p12.1 | <i>MLIP</i> 614106 | Muscular LMNA-interacting protein | Abath Neto et al (2021) | |
| Congenital amyotrophy | 5.50 | AR | | 16p13.3 | <i>CACNA1H</i> 607904 | Calcium channel, voltage-dependent, T type, alpha-1H subunit | Carter et al (2019) | |
| Wieacker-Wolff syndrome | 5.51 | XLR | WRWF 314580 | Xq11.2 | <i>ZC4H2</i> 300897 | Zinc Finger C4H2 domain-containing protein | Hennekam et al (1991) Hirata et al (2013) | |

GROUP 6. MYOTONIC SYNDROMES

| | | | | | | | | |
|--|-----|----|---|----------|--|--|---|---|
| Myotonic dystrophy 1 (Steinert) | 6.1 | AD | DM1 160900 | 19q13.32 | <i>DMPK</i> 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) Ranum et al (1998) Liquori et al (2001) | |
| Myotonic dystrophy type 2 (proximal myotonic myopathy) | 6.2 | AD | DM2 (PROMM) 602668 | 3q21.3 | <i>CNBP</i> (formerly <i>ZNF9</i>) 116955 | CCHC-type Zinc finger nucleic acid-binding protein (Zinc finger protein 9) | | |
| 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 | <i>CAV3</i> 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) |
| Rippling muscle disease, recessive | 6.7 | AR | RMD2 606072 | 3p25.3 | <i>CAV3</i> 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 | <i>HSPG2</i> 14246 | Heparan sulfate proteoglycan of basement membrane (perlecan) | Nicole et al (1995, 2000) | |
| Brody disease | 6.9 | AR | BROD 601003 | 16p11.2 | <i>ATP2A1</i> (formerly <i>SERCA1</i>) 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 | <i>CLCN1</i> 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 | <i>CLCN1</i> 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 | <i>SCN4A</i> 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 CMYP22A, CMYP22B (group 3), HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16) |
| Hypokalaemic periodic Paralysis, type 2 | 7.4 | AD | HOKPP2 613345 | 17q23.3 | <i>SCN4A</i> 603967 | Sodium channel, voltage-gated, type IV, alpha subunit | Bulman et al (1999) Jurkat-Rott et al (2000) | allelic to CMYP22A, CMYP22B (group 3), HYPP, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to <i>SCN4A</i> (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) |
|--|-------------------------|-------------|---|------------|-----------------------------|---|---|---|
| Hypokalaemic periodic paralysis | 7.5 | AD | | 1q23.2 | <i>ATPIA2</i> 182340 | ATPase, NA+/K+ transporting alpha-2 polypeptide | Sampedro et al (2018) | |
| Paramyotonia congenita | 7.6 | AD | PMC 168300 | 17q23.3 | <i>SCN4A</i> 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 CMYP22A, CMYP22B (group 3), HYPP, HOKPP2 and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16) |
| Potassium-aggravated myotonia | 7.7 | AD | 608390 | 17q23.3 | <i>SCN4A</i> 603967 | Sodium channel, voltage-gated, type IV, alpha subunit | Ptáček et al (1992, 1994) Heine et al (1993) Lerche et al (1993) | allelic to CMYP22A, CMYP22B (group 3), HYPP, HOKPP2 and PMC (group 7), CMS16 (group 11), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16) |
| Calcium channel | | | | | | | | |
| Hypokalaemic periodic paralysis, type 1 | 7.8 | AD | HOKPP1 170400 | 1q32.1 | <i>CACNA1S</i> 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 LGMD related to <i>CACNA1S</i> (group 1), CMYP18 (group 3), and MHS5 (group 8) |
| Acetazolamide responsive hereditary paroxysmal cerebellar ataxia (also listed in group 13 "Ataxias") | 7.9 | AD | APCA 108500 | 19p13.13 | <i>CACNA1A</i> 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 | <i>CACNA1A</i> 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 | <i>KCNE3</i> 604433 | Potassium channel, voltage-gated, Isk-related family, member 3 | Abbott et al (2001) | Allelic to BRGDA6 (group 10B) |
| Episodic ataxia/myokymia syndrome | 7.12 | AD | EA1 160120 | 12p13.32 | <i>KCNA1</i> 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 | <i>KCNJ18</i> 613236 | Potassium channel, inwardly rectifying, subfamily J, member 18 (Kir2.6) | Ryan et al (2010) | |
| Periodic paralysis, potassium sensitive cardiodysrhythmic (Andersen's syndrome) | 7.14 | | | | | | <i>see LQ7 under hereditary cardiomyopathies (group 10B, online only)</i> | |
| Long QT syndromes | 7.15 | | | | | | <i>see under hereditary cardiomyopathies (group 10B, online only)</i> | |

GROUP 8. MALIGNANT HYPERTHERMIAS

| | | | | | | | | |
|------------------------|-----|----|----------------|-------------|--------------------------|--|---|--|
| Malignant hyperthermia | 8.1 | AD | MHS1 145600 | 19q13.2 | <i>RYR1</i> 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 <i>RYR1</i> (group 2) CFTD, CNM related to <i>RYR1</i> , CCD, CNMDU1, minicore myopathy with external ophthalmoplegia, DuCD (group 3), late onset axial myopathy (group 5), Fetal akinesia related to <i>RYR1</i> (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 | <i>CACNA1S</i> 114208 | Calcium channel, voltage-dependent, L type, alpha 1S subunit | Monnier et al (1997) | Allelic to LGMD related to <i>CACNA1S</i> (group 1), CMYP18 (group 3), and HOKPP1 (group 7) |

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|--|-------------------------|-------------|---|------------|-----------------------------|--|---|---|
| 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 | 9.1 | AR | GSD2 232300 | 17q25.3 | <i>GAA</i> 606800 | Glucosidase, alpha acid | Hers et al (1963) Martiniuk et al (1990) Wokke et al (1995) | Allelic to Pompe disease, Glycogenosis, generalized, cardiac form (group 10A) |
| Glycogen storage disease type III | 9.2 | AR | GSD3 232400 | 1p21.2 | <i>AGL</i> 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 | <i>GBE1</i> 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 | <i>PYGM</i> 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 | <i>PFKM</i> 610681 | Phosphofruktokinase, muscle type | Tarui et al (1965) Nakajima et al (1991) Howard et al (1996) | |
| Glycogen storage disease type IXd (ex type VIII) | 9.6 | XLR | GSD9D 300559 | Xq13.1 | <i>PHKA1</i> 311870 | Phosphorylase kinase, alpha-1 subunit | Wehner et al (1994) Burwinkel et al (2004) | |
| Congenital disorder of glycosylation, type It | 9.7 | AR | CDG1T 614921 | 1p31.3 | <i>PGM1</i> 171900 | Phosphoglucomutase 1 | Stojkovic et al (2009) | |
| Glycogen storage disease type XV | 9.8 | AR | GSD15 613507 | 3q24 | <i>GYGI</i> 603942 | Glycogenin 1 | Moslemi et al (2010) | allelic to PGBM2 (group 9) |
| Glycogen storage disease type 0 | 9.9 | AR | GSD0B 611556 | 9q13.33 | <i>GYS1</i> 138570 | Glycogen synthase 1 | Kolberg et al (2007) | |
| Glycogen storage disease of heart, lethal congenital | 9.10 | AD | 261740 | 7q36.1 | <i>PRKAG2</i> 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 | <i>RBCK1</i> 610924 | RanBP-type and C3HC4-type zinc finger containing 1 | Nilsson et al (2013) | |
| Polyglucosan Body Myopathy 2 | 9.12 | AR | PGBM2 616199 | 3q24 | <i>GYGI</i> 603942 | Glycogenin 1 | Malfatti et al (2014) | allelic to GSD15 (group 9) |
| Glycolytic pathway | | | | | | | | |
| Phosphoglycerate kinase 1 deficiency | 9.13 | XLR | 300653 | Xq21.1 | <i>PGK1</i> 311800 | Phosphoglycerate kinase 1 | DiMauro et al (1981, 1983) Rosa et al (1982) | |
| Glycogen storage disease type X | 9.14 | AR | GSD10 261670 | 7p13 | <i>PGAM2</i> 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 | <i>LDHA</i> 150000 | Lactate dehydrogenase A | Kanno et al (1980) Scrable et al (1990) | |
| Glycogen storage disease type XIII | 9.16 | AD | GSD13 612932 | 17p13.2 | <i>ENO3</i> 131370 | Enolase 3 | Comi et al (2001) | |
| Erythrocyte lactate transporter defect (Myopathy due to actate transporter defect) | 9.17 | AD | 245340 | 1p13.2 | <i>SLC16A1</i> 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 | AD, AR | 255110 | 1p32.3 | <i>CPT2</i> 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 | <i>SLC22A5</i> 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 | <i>SLC25A20</i> 613698 | Solute carrier family 25 (carnitine/ acylcarnitine translocase), member 20 | Huizing et al (1997) Ogawa et al (2000) | |

| 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) |
|--|-------------------------|-------------|---|---------------|-----------------------------|--|--|--|
| Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIA) | 9.21 | AR | MADD 231680 | 15q24.2-q24.3 | <i>ETF A</i> 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 | <i>ETFB</i> 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 | <i>ETFDH</i> 231675 | Electron-transfer-flavoprotein dehydrogenase | Beard et al (1993) | |
| Acyl-CoA dehydrogenase (very long chain) deficiency | 9.24 | AR | ACADVL D 201475 | 17p13.1 | <i>ACADVL</i> 609575 | Acyl-Coenzyme A dehydrogenase, very long chain | Aoyama (1993, 1995) Strauss et al (1995) Mathur et al (1999) | |
| Mitochondrial complex I deficiency, nuclear type 20 | 9.25 | AR | MC1DN20 611126 | 3q21.3 | <i>ACAD9</i> 611103 | Acyl-CoA dehydrogenase family member 9 (M) | Fragaki et al (2016) | |
| Triglyceride storage disease with ichthyosis (Chanarin-Dorfman syndrome) | 9.26 | AR | CDS 275630 | 3p21.33 | <i>ABHD5</i> 604780 | Abhydrolase domain containing 5 | Lefevre et al (2001) | |
| Neutral lipid storage disease with myopathy without ichthyosis | 9.27 | AR | NLSDM 610717 | 11p15.5 | <i>PNPLA2</i> 609059 | Patatin-like phosphorylase domain-containing protein 2 (Adipose triglyceride lipase) | Fischer et al (2007) | |
| Myoglobinuria, acute recurrent, autosomal recessive | 9.28 | AR | 268200 | 2p25.1 | <i>LPIN1</i> 605518 | lipin 1 (phosphatidic acid phosphatase 1) | Zeharia et al (2008) | |
| Mitochondrial myopathy with lactic acidosis | 9.29 | AR | MMLA 251950 | 7q31.1 | <i>PNPLA8</i> 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 | <i>FLAD1</i> 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 | <i>MYH7</i> 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, CMYP7A, CMYP7B, MSM with eccentric core disease (group 3), MPD1 (group 4), CMD1S and LVNC5 (group 10A) |
| Familial hypertrophic cardiomyopathy, 2 | 10.2 | AD | CMH2 115195 | 1q32.1 | <i>TNNT2</i> 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 | <i>TPM1</i> 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 | <i>MYBPC3</i> 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 | <i>PRKAG2</i> 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 | <i>TNNI3</i> 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 | <i>MYL3</i> 160790 | Myosin, light chain 3, alkali; ventricular, skeletal, slow | Poetter et al (1996) | |
| Familial hypertrophic cardiomyopathy, 9 | 10.8 | AD | CMH9 613765 | 2q31.2 | <i>TTN</i> 188840 | Titin | Satoh et al (1999) | Allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> , MmD related to <i>TTN</i> and CMYP5 (group 3), TMD (group 4), MFM9 (group 5), CMD1G (group 10A), LCCS related to <i>TTN</i> (group 12) |

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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) |
|--|-------------------------|-------------|---|------------|-----------------------------|--|---|---|
| Familial hypertrophic cardiomyopathy, 10 | 10.9 | AD | CMH10 608758 | 12q24.11 | <i>MYL2</i> 160781 | Myosin, light chain 2, regulatory, cardiac, slow | Poetter et al (1996) | Allelic to MF12 (group 3) |
| Familial hypertrophic cardiomyopathy, 11 | 10.10 | AD | CMH11 612098 | 15q14 | <i>ACTC1</i> 102540 | Actin, alpha, cardiac muscle | Mogensen et al (1999) | allelic to CMD1R, LVNC4 (group 10A) |
| Familial hypertrophic cardiomyopathy, 12 | 10.11 | AD | CMH12 612124 | 11p15.1 | <i>CSRP3</i> 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 | <i>TNNC1</i> 191040 | Slow troponin C | Landstrom et al (2008) | allelic to CMD1Z (group 10A) |
| Familial hypertrophic cardiomyopathy, 14 | 10.13 | AD | CMH14 613251 | 14q11.2 | <i>MYH6</i> 160710 | Myosin, heavy chain 6, cardiac muscle, alpha | Carniel et al (2005) | allelic to CMD1EE (group 10A) and SSS3 (group 10B) |
| Hypertrophic cardiomyopathy, 15 | 10.14 | AD | CMH15 613255 | 10q22.2 | <i>VCL</i> 193065 | Vinculin | Vasile et al (2006) | allelic to CMD1W (group 10) |
| Familial hypertrophic cardiomyopathy, 1 | 10.15 | AD, digenic | CMH1 192600 | 20q11.21 | <i>MYLK2</i> 606566 | Myosin light chain kinase 2 | Davis et al (2001) | |
| Familial hypertrophic cardiomyopathy, 1 | 10.16 | AD | CMH1 192600 | 3p25.3 | <i>CAV3</i> 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 | <i>MYOZ2</i> 605602 | Myozenin 2 (calsarcin 1) | Osio et al (2007) | |
| Hypertrophic cardiomyopathy, 17 | 10.18 | AD | CMH17 613873 | 20q13.12 | <i>JPH2</i> 605267 | Junctophilin-2 | Landstrom et al (2007) Matsuhita et al (2007) | |
| Hypertrophic cardiomyopathy, 18 | 10.19 | AD | CMH18 613874 | 6q22.31 | <i>PLN</i> 172405 | Phospholamban | Minamisawa et al (2003) Landstrom et al (2011) | allelic to CMDIP (group 10A) |
| Hypertrophic cardiomyopathy, 19 | 10.20 | AD | CMH19 61387 | 19p13.11 | <i>CALR3</i> 611414 | Calreticulin 3 | Chiu et al (2007) | |
| Hypertrophic cardiomyopathy, 20 | 10.21 | AD | CMH20 613876 | 1p31.1 | <i>NEXN</i> 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 | <i>ANKRD1</i> 609599 | Ankyrin repeat domain-protein 1 | Arimura et al (2009) | allelic to dilated cardiomyopathy (group 10A) |
| Hypertrophic cardiomyopathy, 21 | 10.23 | AD | CMH21 614676 | 7p12.1-q21 | ? | ? | Song et al (2006) | |
| Hypertrophic cardiomyopathy, 22 | 10.24 | AD | CMH22 615248 | 10q21.3 | <i>MYPN</i> 608517 | Myopalladin | Purevjav et al (2012) | allelic to NEM11 (group 3), CMD1KK and RCM4 (group 10A) |
| Hypertrophic cardiomyopathy, 23 | 10.25 | AD | CMH23 612158 | 1q43 | <i>ACTN2</i> 102573 | Actinin alpha-2 | Chiu C et al (2010) | allelic to CMYP8 (group 3), MPD6, distal myopathy with facial weakness (group 4), CMD1AA (group 10A) |
| Hypertrophic cardiomyopathy, 24 | 10.26 | AD | CMH24 601493 | 10q23.2 | <i>LDB3</i> 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 | 10.27 | AD | CMH25 607487 | 17q12 | <i>TCAP</i> 604488 | Titin-cap (telethonin) | Hayashi et al (2004) | allelic to LGMDR7 (group 1), CMD related to <i>TCAP</i> (group 2), CMD1N (group 10A) |
| Hypertrophic cardiomyopathy, 26 | 10.28 | AD | CMH26 617047 | 7q32.1 | <i>FLNC</i> 102565 | Filamin C | Valdes-Mas et al (2014) | allelic to MFM5 (group 5) and MPD4 (group 4) and RCM5 (group 10A) |
| Hypertrophic cardiomyopathy, 27 | 10.29 | AR | CMH27 618052 | 15q25.3 | <i>ALPK3</i> 617608 | Alpha Kinase 3 | Almmani et al (2016) | |
| Hypertrophic cardiomyopathy, 28 | 10.30 | AD | CMH28 619402 | 18q12.2 | <i>FHOD3</i> 609691 | Formin homology-2 domain-containing protein 3 | Ochoa et al (2018) | |
| Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies | 10.31 | AR | CMH29 620236 | 3q27.1 | KLHL24 611295 | Kelch-like 24 | Hedberg-Oldfors et al (2019) | |
| Mitochondrial complex I deficiency, nuclear type 11 | 10.32 | AR | MC1DN11 618234 | 15q15.1 | <i>NDUFA1</i> 606934 | NADH dehydrogenase (ubiquinone) complex I, Assembly factor 1 (M) | Fassone et al (2011) | |
| Combined oxydative phosphorylation deficiency 3 | 10.33 | AR | COXPD3 610505 | 12q14.1 | <i>TSM</i> 604723 | Ts translation elongation factor, mitochondrial (M) | Smeitink et al (2006) | |

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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) |
|---|-------------------------|-------------|---|--------------|-----------------------------|---|--|---|
| Combined oxydative phosphorylation deficiency 8 | 10.34 | AR | COXPD8 614096 | 6p21.1 | <i>AARS2</i> 612035 | Alanyl-tRNA synthetase 2, mitochondrial (M) | Götz et al (2011) | |
| Combined oxydative phosphorylation deficiency 9 | 10.35 | AR | COXPD9 614582 | 3q21.1 | <i>MRPL3</i> 607118 | Mitochondrial ribosomal protein L3 (M) | Galmiche et al (2011) | |
| Combined oxydative phosphorylation deficiency 10 | 10.36 | AR | COXPD10 614702 | 6q13 | <i>MTO1</i> 614667 | Mitochondrial tRNA translation optimization 1 (M) | Ghezzi et al (2012) Baruffini et al (2013) | |
| Combined oxydative phosphorylation deficiency 16 | 10.37 | AR | COXPD16 615395 | 2q36.1 | <i>MRPL44</i> 611849 | Mitochondrial ribosomal protein L44 (M) | Carroll et al (2013) | |
| Mitochondrial complex IV deficiency, nuclear type 2 | 10.38 | AR | MC4DN2 (CEMCOX1) 604377 | 22q13.33 | <i>SCO2</i> 604272 | Cytochrome c oxidase assembly protein (M) | Papadopoulou et al (1999) | allelic to CMT2 related to SCO2 (group 14) |
| Mitochondrial complex IV deficiency, nuclear type 6 | 10.39 | AR | MC4DN6 (CEMCOX2) 615119 | 10q24.2 | <i>COX15</i> 603646 | Cytochrome c oxidase assembly factor Cox15 (M) | Antonicka et al (2003) | |
| Dilated cardiomyopathies | | | | | | | | |
| Dilated cardiomyopathy, 1A | 10.40 | AD | CMD1A 115200 | 1q22 | <i>LMNA</i> 150330 | Lamin A/C | Fatkin et al (1999) | Allelic to EDMD2, EDMD3 (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 | 10.41 | AD | CMD1B 600884 | 9q13 | ? | ? | Krajcinovic et al (1995) | |
| Dilated cardiomyopathy, 1C, with or without LVNC | 10.42 | AD | CMD1C 601493 | 10q23.2 | <i>LDB3</i> 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 | 10.43 | AD | CMD1D 601494 | 1q32.1 | <i>TNNT2</i> 191045 | Troponin type 2 (cardiac) | Durand et al (1995) Kamisago et al (2000) | allelic to CMH2, RCM3 and and LVNC6 (group 10A) |
| Dilated cardiomyopathy, 1E | 10.44 | AD | CMD1E 601154 | 3p22.2 | <i>SCN5A</i> 600163 | Sodium channel, voltage-gated, type V, alpha | McNair et al (2004) | allelic to BRGDA1, ATFB10, LQT3, SSS1 (group 10B) |
| Dilated cardiomyopathy, 1G | 10.45 | AD | CMD1G 604145 | 2q31.2 | <i>TTN</i> 188840 | Titin | Siu et al (1999) Gerull et al (2002) Itoh-Satoh et al (2002) | Allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> , MmD related to <i>TTN</i> and CMYP5 (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), LCCS related to <i>TTN</i> (group 12) |
| Dilated cardiomyopathy, 1H | 10.46 | AD | CMD1H 604288 | 2q14-q22 | ? | ? | Jung et al (1999) | |
| Dilated cardiomyopathy, 1I | 10.47 | AD | CMD1I 604765 | 2q35 | <i>DES</i> 125660 | Desmin | Li et al (1999) | Allelic to MFM1 with or without ARCV7 (group 5), ARVC7 (group 10B) and myopathy, cardiomyopathy and CMS (group 11) |
| Dilated cardiomyopathy, 1J | 10.48 | AD | CMD1J 605362 | 6q23.2 | <i>EYA4</i> 603550 | Eyes absent homolog | Schönberger et al (2005) | |
| Dilated cardiomyopathy, 1K | 10.49 | AD | CMD1K 605582 | 6q12-q16 | ? | ? | Sylvius et al (2001) | |
| Dilated cardiomyopathy, 1L | 10.50 | AD | CMD1L 606685 | 5q33.2-q33.3 | <i>SGCD</i> 601411 | Sarcoglycan, delta | Tsubata et al (2000) | allelic to LGMDR6 (group 1) |
| Dilated cardiomyopathy, 1M | 10.51 | AD | CMD1M 607482 | 11p15.1 | <i>CSRFP3</i> 600824 | Cysteine and glycine-rich protein 3 (cardiac LIM protein) | Knoll et al (2002) | allelic to CMH12 (group 10A) |
| Dilated cardiomyopathy, 1N | 10.52 | AD | CMD1N | 17q12 | <i>TCAP</i> 604488 | Titin-cap (Telethonin) | Knoll et al (2002) | allelic to LGMDR7 (group 1), CMD related to <i>TCAP</i> (group 2), CMH25 (group 10A) |
| Dilated cardiomyopathy, 1O | 10.53 | AD | CMD1O 605569 | 12p12.1 | <i>ABCC9</i> 601439 | ATP-binding cassette, subfamily C, member 9 | Bienengraeber et al (2004) | allelic to ATFB12 (group 10B) |
| Dilated cardiomyopathy, 1P | 10.54 | AD | CMD1P 609909 | 6q22.31 | <i>PLN</i> 172405 | phospholamban | Schmitt et al (2003) Haghighi et al (2003, 2006) | allelic to CMH18 (group 10A) |
| Dilated cardiomyopathy, 1Q | 10.55 | AD | CMD1Q 609915 | 7q22.3-q31.1 | ? | ? | Schonberger et al (2005) | |

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| DISEASE NAME | Item line in this group | Inheritance | Locus or disease symbol and OMIM number | Chromosome | Gene symbol and OMIM number | Protein (mitochondrial proteins indicated by symbol [M]) | Key references | Other allelic disease(s) (group in this table) |
|---|-------------------------|-------------|---|------------|-----------------------------|--|---|--|
| Dilated cardiomyopathy, 1R | 10.56 | AD | CMD1R 613424 | 15q14 | <i>ACTC1</i> 102540 | actin, alpha, cardiac muscle | Olson et al (1998) Mogensen et al (1999) | allelic to CMH11, LVNC4 (group 10A) |
| Dilated cardiomyopathy, 1S | 10.57 | AD | CMD1S 613426 | 14q11.2 | <i>MYH7</i> 160760 | Myosin heavy chain 7 (beta), cardiac muscle | Kamisago et al (2000) | Allelic to CFTD, CMYP7A, CMYP7B, MSM with eccentric core disease (group 3), MPD1 (group 4), CMH1 and LVNC5 (group 10A) |
| Dilated cardiomyopathy, 1T | 10.58 | AD | CMD1T | 12q23.1 | <i>TMPO</i> 188380 | Thymopoietin (lamina-associated polypeptide 2) | Taylor et al (2005) | |
| Dilated cardiomyopathy, 1U | 10.59 | AD | CMD1U 613694 | 14q24.2 | <i>PSEN1</i> 104311 | Presenilin 1 | Li et al (2006) | |
| Dilated cardiomyopathy, 1V | 10.60 | AD | CMD1V 613697 | 1q42.13 | <i>PSEN2</i> 600759 | Presenilin 2 | Li et al (2006) | |
| Dilated cardiomyopathy, 1W | 10.61 | AD | CMD1W 611407 | 10q22.2 | <i>VCL</i> 193065 | Vinculin | Olson et al (2002) Vasile et al (2006) | allelic to CMH15 (group 10) |
| Dilated cardiomyopathy related, 1X | 10.62 | AR | CMD1X 611615 | 9q31.2 | <i>FKTN</i> 607440 | Fukutin | Murakami et al (2006) | allelic to MDDGC4 (group 1), MDDGA4 and MDDGB4 (group 2) |
| Dilated cardiomyopathy, 1Y | 10.63 | AD | CMD1Y 611878 | 15q22.2 | <i>TPM1</i> 191010 | Tropomyosin-1 | Olson et al (2010) | allelic to CMH3 and LVNC9 (group 10A) |
| Dilated cardiomyopathy, 1Z | 10.64 | AD | CMD1Z 611879 | 3p21.1 | <i>TNNC1</i> 191040 | Slow troponin C | Mogensen et al (2004) | allelic to CMH13 (group 10A) |
| Dilated cardiomyopathy, 1AA, with or without LVNC | 10.65 | AD | CMD1AA 612158 | 1q42-q43 | <i>ACTN2</i> 102573 | Actinin alpha-2 | Mohapatra et al (2003) | allelic to CMYP8 (group 3), MPD6 (group 4), CMH23 (group 10A) |
| Dilated cardiomyopathy, 1BB | 10.66 | AD, AR | CMD1BB 612877 | 18q12.1 | <i>DSG2</i> 125671 | Desmoglein 2 | Posch et al (2008) | allelic to ARVD10 (group 10B) |
| Dilated cardiomyopathy, 1CC | 10.67 | AD | CMD1CC 613122 | 1p31.1 | <i>NEXN</i> 613121 | Nexilin F-actin binding protein | Hassel et al (2009) | allelic to CMH20 (group 10A) |
| Dilated cardiomyopathy, 1DD | 10.68 | AD | CMD1DD 613172 | 10q25.2 | <i>RBM20</i> 613171 | RNA-binding motif protein 20 | Brauch et al (2009) | |
| Dilated cardiomyopathy, 1EE | 10.69 | AD | CMD1EE 613252 | 14q11.2 | <i>MYH6</i> 160710 | Myosin, heavy chain 6, cardiac muscle, alpha | Carniel et al (2005) | allelic to CMH14 (group 10A), SSS3 (group 10B) |
| Dilated cardiomyopathy, 1FF | 10.70 | AD | CMD1FF 613286 | 19q13.42 | <i>TNNI3</i> 191044 | Cardiac troponin I | Carballo et al (2009) | allelic to CMH7, CDM2A, RCM1 (group 10A) |
| Dilated cardiomyopathy, 1GG | 10.71 | AR | CMD1GG 613642 | 5p15.33 | <i>SDHA</i> 600857 | Succinate dehydrogenase complex, subunit a, flavoprotein (M) | Levitas et al (2010) | |
| Dilated cardiomyopathy, 1HH | 10.72 | AD | CMD1HH 613881 | 10q26.11 | <i>BAG3</i> 603883 | Bcl2-associated athanogene 3 | Norton et al (2011) | allelic to MFM6 (group 5) and CMT related to BAG3 (group 14) |
| Dilated cardiomyopathy, 1II | 10.73 | AD | CMD1II 615184 | 11q23.1 | <i>CRYAB</i> 123590 | Alpha-B crystallin | Inagaki et al (2006) | Allelic to MFM2 (group 5) and CMT2 related to <i>CRYAB</i> (group 14) |
| Dilated cardiomyopathy, 1JJ | 10.74 | AD | CMD1JJ 615235 | 6q21 | <i>LAMA4</i> 600133 | Laminin, alpha-4 | Knöll et al (2007) | |
| Dilated cardiomyopathy, 1KK | 10.75 | AD | CMD1KK 615248 | 10q21.3 | <i>MYPN</i> 608517 | Myopalladin | Duboscq-Bidot (2008) | allelic to NEM11 (group 3), CMH22 and RCM4 (group 10A) |
| Dilated cardiomyopathy, 1LL | 10.76 | AD | CMD1LL 615373 | 1p36.32 | <i>PRDM16</i> 605557 | PR domain-containing protein 16 | Arndt et al (2013) | allelic to LVNC8 (group 10A) |
| Dilated cardiomyopathy, 1MM | 10.77 | AD | CMD1MM 615396 | 11p11.2 | <i>MYBPC3</i> 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 | 10.78 | AD | CMD1NN 615916 | 3p25.2 | <i>RAF1</i> 164760 | V-Raf-1 murine leukemia viral oncogene homolog 1 | Dhandapany et al (2014) | |
| Dilated cardiomyopathy, 1OO | 10.79 | AD | CMD1OO 620247 | 17q22 | <i>VEZFI</i> 606747 | Vascular endothelial zing finger 1 | Shi et al (2023) | |
| Dilated cardiomyopathy related to <i>ILK</i> | 10.80 | AD | | 11p15.4 | <i>ILK</i> 602366 | Integrin-linked kinase | Knöll et al (2007) | |
| Dilated cardiomyopathy related to <i>ANKRD1</i> | 10.81 | AD | | 10q23.31 | <i>ANKRD1</i> 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 | 10.82 | AR | CMD2A 611880 | 19q13.42 | <i>TNNI3</i> 191044 | Cardiac troponin I | Carballo et al (2009) | allelic to CMH7, CMD1FF, RCM1 (group 10A) |

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| DISEASE NAME | Item line in this group | Inheritance | Locus or disease symbol and OMIM number | Chromosome | Gene symbol and OMIM number | Protein (mitochondrial proteins indicated by symbol [M]) | Key references | Other allelic disease(s) (group in this table) |
|---|-------------------------|-------------|---|--------------|-----------------------------|--|---|---|
| Dilated cardiomyopathy, 2B | 10.83 | AR | CMD2B 614672 | 7q21.2 | GATAD1 614518 | GATA zinc finger domain-containing protein 1 | Theis et al (2011) | |
| Dilated cardiomyopathy, 2C | 10.84 | AR | CDMD2C 618189 | 1p34.2 | PPCS 609853 | Phosphopanto-thenosylcystein synthetase | Iuso et al (2018) | |
| Dilated cardiomyopathy, 2D | 10.85 | AR | CMD2D 619371 | 16p13.3 | RPL3L 617416 | Ribosomal protein L3-like | Ganapathi et al (2020) | |
| Dilated cardiomyopathy, 2E | 10.86 | AR | CMD2E 619492 | 20q13.12 | JPH2 605267 | Junctophilin-2 | Vasilescu et al (2018) Jones et al (2019) | Allelic to CMH17 (group 10A) |
| Dilated cardiomyopathy, 2F | 10.87 | AR | CMD2F 619747 | 14q32.33 | BAG5 603885 | Bag cochaperone | Hakui et al (2022) | |
| Dilated cardiomyopathy, 2G | 10.88 | AR | CMD2G 619897 | 7q31.32 | LMOD2 68006 | Leiomodoin 2 | Ahrens-Nicklas et al (2019) | |
| Dilated cardiomyopathy, 2H | 10.89 | AR | CMD2H 620203 | 19p13.13 | GET3 601913 | Guided entry of tail-anchored proteins factor 3, ATPase | Verhagen et al (2019) | |
| Dilated cardiomyopathy, 2I | 10.90 | AR | CMD2I 620462 | 6p22.3 | CAP2 618385 | Cyclase associated actin cytoskeleton regulatory protein 2 | Aspit et al (2019) | |
| Dilated cardiomyopathy, 3A | 10.91 | XLR | CMD3A | Xq28 | TAZ 300394 | Tafazzin | Gedeon et al (1995) | allelic to BTHS (group 10A) |
| Dilated cardiomyopathy, 3B | 10.92 | XLR | 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 <i>SYNE1</i> | 10.93 | 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 <i>CAVIN4</i> | 10.94 | AD | | 9q31.1 | CAVIN4 617714 | Caveolae-associated protein 4 (MURC) | Rodriguez et al (2011) | |
| Dilated cardiomyopathy related to <i>DOLK</i> | 10.95 | AR | CDGM1 610768 | 9q34.11 | DOLK 610746 | Dolichol kinase | Kranz et al (2007) Lefeber et al (2011) | |
| Restrictive cardiomyopathies | | | | | | | | |
| Restrictive cardiomyopathy, 1 | 10.96 | AD | RCM1 115210 | 19q13.42 | TNNI3 191044 | Cardiac troponin I | Mogensen et al (2003) | allelic to CMH7, CMD1FF, CMD2A (group 10A) |
| Restrictive cardiomyopathy, 2 | 10.97 | | RCM2 609578 | 10q23.3 | ? | ? | Zhang et al (2005) | |
| Restrictive cardiomyopathy, 3 | 10.98 | 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 | 10.99 | 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 | 10.100 | 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) |
| Restrictive cardiomyopathy, 6 | 10.101 | AR | RCM6 619433 | 5q31.2 | KIF20A 605664 | Kinesin family member 20A | Louw et al (2018) | |
| Other non arrhythmic hereditary cardiomyopathies | | | | | | | | |
| Pompe disease, Glycogenosis, generalized, cardiac form | 10.102 | AR | GSD2 232300 | 17q25.3 | GAA 606800 | Glucosidase, alpha; acid | Hers (1963) Martiniuk et al (1990) Wokke et al (1995) | Allelic to Pompe disease (group 9) |
| Barth syndrome | 10.103 | XLR | BTHS 302060 | Xq28 | TAZ 300394 | Tafazzin | Bolhuis et al (1991) Bione et al (1996) | allelic to CMD3A (group 10A) |
| Left ventricular noncompaction, 1 | 10.104 | AD | LVNC1 604169 | 18q12.1 | DTNA 601239 | Dystrobrevin, alpha | Ichida et al (2001) | Allelic to LGMD related to <i>DTNA</i> (group 1) |
| Left ventricular noncompaction, 2 | 10.105 | | LVNC2 609470 | 11q15 | ? | ? | Sasse-Klaassen et al (2004) | |
| Left ventricular noncompaction, 3 | 10.106 | AD | LVNC3 601493 | 10q23.2 | LDB3 605906 | LIM domain binding-3 | Vatta et al (2003) | allelic to Late onset distal myopathy (group 4), MFM4 (group 5), CMH24 and CMD1C (group 10A) |
| Left ventricular noncompaction, 4 | 10.107 | AD | LVNC4 613424 | 15q14 | ACTC1 102540 | Alpha-cardiac actin | Monserat et al (2007) | allelic to CMH11, CMD1R (group 10A) |
| Left ventricular noncompaction, 5 | 10.108 | AD | LVNC5 613426 | 14q11.2 | MYH7 160760 | Myosin heavy chain 7 (beta), cardiac muscle | Klaassen et al (2008) | Allelic to CFTD, CMYP7A, CMYP7B, MSM with eccentric core disease (group 3), MPD1 (group 4), CMH1, CMD1S (group 10A) |

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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) |
|------------------------------------|-------------------------|-------------|---|------------|-----------------------------|--|--|---|
| Left ventricular noncompaction, 6 | 10.109 | AD | LVNC6 601494 | 1q32.1 | <i>TNNT2</i> 191045 | Cardiac Troponin T | Luedde et al (2010) | allelic to CMH2, CMD1D and RCM3 (group 10A) |
| Left ventricular noncompaction, 7 | 10.110 | AD | LVNC7 615092 | 18q11.2 | <i>MIB1</i> 608677 | Mindbomb, drosophila, homolog of | Luxan et al. (2013) | |
| Left ventricular noncompaction, 8 | 10.111 | AD | LVNC8 615373 | 1p36.32 | <i>PRDM16</i> 605557 | PR domain-containing protein 16 | Arndt et al. (2013) | allelic to CMD1LL (group 10A) |
| Left ventricular noncompaction, 9 | 10.112 | AD | LVNC9 611878 | 15q22.2 | <i>TPM1</i> 191010 | Tropomyosin-1 | Probst et al. (2011) | allelic to CMH3, CMD1Y (group 10A) |
| Left ventricular noncompaction, 10 | 10.113 | AD | LVNC10 615396 | 11p11.2 | <i>MYBPC3</i> 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 | 10.114 | XLR | CVDPX 314400 | Xq28 | <i>FLNA</i> 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 | <i>CHRNA1</i> 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 | <i>CHRNBI</i> 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 | <i>CHRNA1</i> 100690 | Cholinergic receptor, nicotinic, alpha 1 | Gomez et al (2002) | allelic to CMS3B, CMS3C (group 11) |
| Myasthenic syndrome, congenital, 4A, Slow-channel | 11.4 | AD, AR | CMS4A 605809 | 17p13.2 | <i>CHRNA1</i> 100690 | Cholinergic receptor, nicotinic, alpha 1 | Ohno et al (1995) Gomez et al (1995) Engel et al (1996) Croxen et al (2002) | allelic to CMS4B, CMS4C (group 11) |
| Myasthenic syndrome, congenital, 1B, Fast-channel | 11.5 | AR | CMS1B 608930 | 2q31.1 | <i>CHRNA1</i> 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 | <i>CHRNBI</i> 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 | <i>CHRNA1</i> 100690 | Cholinergic receptor, nicotinic, alpha 1 | Brownlow et al (2001) | allelic to CMS3A, CMS3C (group 11) |
| Myasthenic syndrome, congenital, 4B, Fast-channel | 11.8 | AR | CMS4B 616324 | 17p13.2 | <i>CHRNA1</i> 100690 | Cholinergic receptor, nicotinic, alpha 1 | 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 | <i>CHRNBI</i> 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 | <i>CHRNA1</i> 100690 | Cholinergic receptor, nicotinic, alpha 1 | Shen et al (2002) | allelic to CMS3A, CMS3B (group 11) |
| Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency | 11.11 | AR | CMS4C 608931 | 17p13.2 | <i>CHRNA1</i> 100690 | Cholinergic receptor, nicotinic, alpha 1 | 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 | <i>COLQ</i> 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 | <i>CHAT</i> 118490 | Choline acetyltransferase | Ohno et al (2001) Maselli et al (2003) | |
| Myasthenic syndrome, congenital, 7, presynaptic and distal motor neuropathy, autosomal dominant | 11.14 | AD | CMS7A 616040 | 1q32.1 | <i>SYT2</i> 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, autosomal recessive | 11.15 | AR | CMS7B 619461 | 1q32.1 | <i>SYT2</i> 600104 | Synaptotagmin 2 | Masseli et al (2020) | Allelic to CMS7 (group 11) and to Distal motor neuropathy related to SYT2 (group 12) |

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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) |
|--|-------------------------|-------------|---|------------|-----------------------------|---|---|--|
| Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects | 11.16 | AR | CMS8 615120 | 1p36.33 | <i>AGRN</i> 103320 | Agrin | Huzé et al (2009) | Allelic to FADS related to <i>AGRN</i> (group 16) |
| Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency | 11.17 | AR | CMS9 616325 | 9q31.3 | <i>MUSK</i> 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 | <i>DOK7</i> 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 | <i>RAPSN</i> 601592 | Receptor-associated protein of the synapse, 43kD (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 | <i>GFPT1</i> 138292 | Glutamine:fructose-6-phosphate amido transterase 1 | Senderek et al (2011) | |
| Myasthenic syndrome, congenital, 13, with tubular aggregates | 11.21 | AR | CMS13 614750 | 11q23.3 | <i>DPAGT1</i> 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 | <i>ALG2</i> 607905 | S. Cerevisiae 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 | <i>ALG14</i> 612866 | S. Cerevisiae homolog of ALG14 (UDP-N-acetylglucosaminyltransferase subunit) | Cossins et al (2013) | |
| Myasthenic syndrome, congenital, 16 | 11.24 | AR | CMS16 614198 | 17q23.3 | <i>SCN4A</i> 603967 | Sodium channel, voltage-gated, type IV, alpha subunit | Tsujino et al (2003) | allelic to CMYP22A, CMYP22B (group 3), HOKPP2, HYPP, PMC and K-aggravated myotonia (group 7), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16) |
| Myasthenic syndrome, congenital, 17 | 11.25 | AR | CMS17 616304 | 11p11.2 | <i>LRP4</i> 604270 | LDL receptor-related protein 4 | Ohkawara et al (2014) | |
| Myasthenic syndrome, congenital, 18 | 11.26 | AD | CMS18 616330 | 20p12.2 | <i>SNAP25</i> 600322 | Synaptosomal associated protein 25 | Shen et al (2014) | |
| Myasthenic syndrome, congenital, 19 | 11.27 | AR | CMS19 616720 | 10q22.1 | <i>COL13A1</i> 120350 | Collagen type XIII alpha 1 chain | Logan et al (2015) | |
| Myasthenic syndrome, congenital, 20, presynaptic | 11.28 | AR | CMS20 617143 | 2q12.3 | <i>SLC5A7</i> 608761 | Solute carrier family 5 (choline transporter) member 7 | Bauche et al (2016) | allelic to HMND7 (group 12) |
| Myasthenic syndrome, congenital, 21, presynaptic | 11.29 | AR | CMS21 617239 | 10q11.23 | <i>SLC18A3</i> 600336 | Solute carrier family 18 (vesicular acetylcholine), member 3 | O'Grady et al (2016) | |
| Myasthenic syndrome, congenital, 22 | 11.30 | AR | CMS22 616224 | 2p21 | <i>PREPL</i> 609557 | Prolyl endopeptidase-like | Regal et al (2014) | |
| Myasthenic syndrome, congenital, 23, presynaptic | 11.31 | AR | CMS23 618197 | 22q11.21 | <i>SLC25A1</i> 190315 | Solute carrier family 25 (mitochondrial carrier, citrate transporter), member 1 | Chaouch et al (2014) | |
| Myasthenic syndrome, congenital, 24, presynaptic | 11.32 | AR | CMS24 618198 | 15q23 | <i>MYO9A</i> 604875 | Myosin IXA | O'Connor et al (2016) | |
| Myasthenic syndrome, congenital, 25 | 11.33 | AR | CMS25 618323 | 12p13.31 | <i>VAMP1</i> 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 | <i>LAMB2</i> 150325 | Laminin-beta 2 | Maselli et al (2009) | |
| Escobar syndrome (multiple pterygium syndrome) | 11.35 | AR | EVMP5 265000 | 2q37.1 | <i>CHRNA3</i> 100730 | Cholinergic receptor, nicotinic, gamma | Hoffman et al (2006) Morgan et al (2006) | |
| Myasthenic syndrome, with plectin defect | 11.36 | AR | | 8q24.3 | <i>PLEC</i> 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 GMPBB | 11.37 | AR | MDDGC14 615352 | 3q21.31 | <i>GMPBB</i> 615320 | GDP-mannose pyrophosphorylase B | Belaya et al (2015) | allelic to LGMDR19/MDDGC14 (group 1); MDDGA14 and MDDGB14 (group 2) |

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|---|-------------------------|-------------|---|------------|--|---|---|---|
| Presynaptic congenital myasthenic syndrome | 11.38 | AR | | 20q13.33 | LAMA5 601033 | Laminin, alpha-5 | Maselli et al (2017) | |
| Presynaptic congenital myasthenic syndrome related to <i>MUNC13-1</i> | 11.39 | AR | | 19p13.11 | UNC13A 609894 | C. Elegans, homolog of UNC13A (MUNC13) | Engel et al (2016) | |
| Congenital myasthenic syndrome related to <i>RPH3A</i> , presynaptic | 11.40 | AR | | 12q24.13 | RPH3A 612159 | Rabphilin 3A | Maselli et al (2018) | |
| Congenital myasthenic syndrome | 11.41 | AR | | 1q25.2 | TORIAIPI (= <i>LAP1B</i>) 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) | |
| Myasthenic syndrome, congenital associated with acetylcholine receptor deficiency | 11.43 | AR | | 2q31.1 | CHRNA1 100690 | Cholinergic receptor, nicotinic, alpha 1 | Abath Neto et al (2017) | allelic to CMS1A and CMS1B (group 11) |
| Myopathy, cardiomyopathy and congenital myasthenic syndrome | 11.44 | AR | | 2q35 | DES 125660 | Desmin | Durmus et al (2016) | allelic to MFM1 with or without ARCV7 (group 5), CMD11 (group 10A), ARVC7 (group 10B) |

GROUP 12. SPINAL MUSCULAR ATROPHIES MOTONEURON DISEASES*Spinal muscular atrophy related to SMNI*

| | | | | | | | | |
|--|------|----|----------------|--------|-----------------------|----------------------------|--|--|
| Spinal muscular atrophy, type I (Werdnig-Hoffman) | 12.1 | AR | SMA1 253300 | 5q13.2 | SMNI 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 | SMNI 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 | SMNI 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 | SMNI 600354 | Survival of motor neuron 1 | Brahe et al (1995) Clermont et al (1995) | allelic to SMA2, SMA3, SMA1 (group 12) |

Proximal spinal muscular atrophy

| | | | | | | | | |
|---|-------|----|------------------|----------|-------------------------|--|---|---|
| Spinal muscular atrophy with congenital bone fractures 1 | 12.5 | 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 | 12.6 | AR | SMABF2 616867 | 10q22.1 | ASCC1 614215 | Activating signal cointegrator 1 complex subunit 1 | Knierim et al (2016) | allelic to Arthrogyrosis related to <i>ASCC1</i> (group 16) |
| Spinal muscular atrophy with progressive myoclonic epilepsy | 12.7 | AR | SMAPME 159950 | 8p22 | ASAH1 613468 | N-acylsphingosine amidohydrolase 1 | Zhou et al (2012) | allelic to FRBRL (#228000) |
| Spinal muscular atrophy and cerebellar hypoplasia | 12.8 | AR | PCH1C 616081 | 13q13.3 | EXOSC8 606019 | Exosome component 8 | Boczonadi et al (2014) | |
| Pontocerebellar hypoplasia, type 1D | 12.9 | AR | PCH1D 618065 | 4q27 | EXOSC9 606180 | Exosome component 9 | Burns et al (2018) | |
| Spinal Muscular Atrophy type IV related to <i>CAPN1</i> | 12.10 | AR | | 11q13.1 | CAPN1 114220 | Calpain 1 | Perez-Siles et al (2022) | Allelic to SPG76 (group 15) |
| Scapuloperoneal spinal muscular atrophy | 12.11 | 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 HMND8 (group 12), HSMN2C (group 14) |
| Spinal motor neuropathy | 12.12 | | | 11q23.2 | RBM7 612413 | RNA-binding motif protein 7 | Giunta et al (2016) | |

Distal hereditary motor neuropathy, recessive

| | | | | | | | | |
|--|-------|----|-----------------|---------|--------------------------|-------------------------------------|-----------------------------|-----------------------------|
| Neuropathy, distal hereditary motor, autosomal recessive 1 | 12.13 | AR | HMNR1 604320 | 11q13.3 | IGHMBP2 600502 | Immunoglobulin Mu-binding protein 2 | Grohmann et al (1999, 2001) | allelic to CMT2S (group 14) |
|--|-------|----|-----------------|---------|--------------------------|-------------------------------------|-----------------------------|-----------------------------|

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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) |
|---|-------------------------|-------------|---|------------|-----------------------------|--|---|--|
| Neuropathy, distal hereditary motor, autosomal recessive 2 | 12.14 | AR | HMNR2 605726 | 9p13.3 | <i>SIGMAR1</i> 601978 | Sigma non-opioid intracellular receptor 1 | Christodoulou et al (2000) Li et al (2015) | allelic to ALS16 (group 12) |
| Neuropathy, distal hereditary motor, autosomal recessive 3 | 12.15 | AR | HMNR3 607088 | 11q13 | ? | ? | Viollet et al (2004) | |
| Neuropathy, distal hereditary motor, autosomal recessive 4 | 12.16 | AR | HMNR4 611067 | 1p36.31 | <i>PLEKHG5</i> 611101 | Pleckstrin homology domain and RhoGEF domain-containing protein G5 | Maystadt et al (2006, 2007) | allelic to CMTRIC (group 14) |
| Neuropathy, distal hereditary motor, autosomal recessive 5 | 12.17 | AR | HMNR5 614881 | 2q35 | <i>DNAJB2</i> 604139 | DnaJ/Hsp40 homolog, subfamily B, member 2 | Blumen et al (2012) | Allelic to CMT related to DNAJB2 (group 14) |
| Neuropathy, distal hereditary motor, autosomal recessive 6 | 12.18 | AR | HMNR6 620011 | 2p11.2 | <i>REEP1</i> 609139 | Receptor expression-enhancing protein 1 | Schottmann et al (2015) | Allelic to HMND12 (group 12) and SPG31 (group 15) |
| Neuropathy, hereditary motor, autosomal recessive 7 | 12.19 | AR | HMNR7 619216 | 1p36.33 | <i>VWAI</i> 611901 | Von Willebrand factor A domain-containing protein 1 | Deschauer et al (2021) Pagnamenta et al (2021) | |
| Neuropathy, hereditary motor, autosomal recessive 8 | 12.20 | AR | HMNR8 618912 | 15q21.1 | <i>SORD</i> 182500 | Sorbitol dehydrogenase | Cortese et al (2020) | Allelic to SORDD (group 14) |
| Neuropathy, distal hereditary motor, autosomal recessive 9 | 12.21 | AR | HMNR9 620402 | 16q12.3 | <i>COQ7</i> 601683 | Coenzyme Q7 | Jacquier et al (2023) Liu et al (2023) Smith et al (2023) | Allelic to COQ10D8 (group 16) |
| Neuropathy, distal hereditary motor, autosomal recessive | 12.22 | AR | HMNR10 620542 | 14q32.2 | <i>VRK1</i> 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) |
| Neuropathy, Distal hereditary motor related to <i>HINT1</i> | 12.23 | AR | | 5q23.3 | <i>HINT1</i> 601314 | Histidine triad nucleotide-binding protein | Zhao et al (2014) | Allelic to NMAN (group 14) |
| Distal hereditary motor neuropathy, dominant | | | | | | | | |
| Neuropathy, distal hereditary motor, type 1 | 12.24 | AD | HMND1 182960 | 7q34-q36 | ? | ? | Gopinath et al (2007) | |
| Neuropathy, distal hereditary motor, autosomal dominant 2 | 12.25 | AD | HMND2 158590 | 12q24.23 | <i>HSPB8</i> 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) |
| Neuropathy, distal hereditary motor, autosomal dominant 3 | 12.26 | AD | HMND3 608634 | 7q11.23 | <i>HSPB1</i> 602195 | Heat-shock 27-kD protein 1 | Evgrafov et al (2004) | allelic to CMT2F (group 14) |
| Neuropathy, distal hereditary motor, autosomal dominant 4 | 12.27 | AD | HMND4 613376 | 5q11.2 | <i>HSPB3</i> 604624 | Heat shock 27-kD protein 3 | Kolb et al. (2010) | |
| Neuropathy, distal hereditary motor, autosomal dominant 5 | 12.28 | AD | HMND5 600794 | 7p14.3 | <i>GARS1</i> 600287 | Glycyl-tRNA synthetase | Christodoulou et al (1995) Antonellis et al (2003) | allelic to CMT2D (group 14) |
| Neuropathy, distal hereditary motor, autosomal dominant 6 | 12.29 | AD | HMND6 615575 | 5q32 | <i>FBXO38</i> 608533 | F-box only protein 38 | Sumner et al (2013) | |
| Neuropathy, distal hereditary motor, autosomal dominant 7 | 12.30 | AD | HMND7 158580 | 2q12.3 | <i>SLC5A7</i> 608761 | Solute carrier family 5 (choline cotransporter), member 7 | McEntagart et al. (2001) Barwick et al (2012) | allelic to CMS20 (group 11) |
| Neuropathy, distal hereditary motor, autosomal dominant 8 | 12.31 | AD | HMND8 600175 | 12q24.11 | <i>TRPV4</i> 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) |
| Neuropathy, distal hereditary motor, autosomal dominant 9 | 12.32 | AD | HMND9 61772 | 14q32.2 | <i>WARS1</i> 191050 | Tryptophanyl-tRNA synthetase 1 | Tsai et al (2017) | |
| Neuropathy, distal hereditary motor, autosomal dominant 10 | 12.33 | AD | HMND10 620080 | 2p23.3 | <i>EMILIN1</i> 130660 | Elastin microfibril interfacier 1 | Capuano et al (2016) | |
| Neuropathy, distal hereditary motor, autosomal dominant 11 | 12.34 | AD | HMND11 620528 | 9q34.11 | <i>SPTAN1</i> 182810 | Spectrin, alpha, nonerythrocytic 1 | Beijer 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) |
|--|-------------------------|-------------|---|--------------|-----------------------------|--|---|--|
| Neuronopathy, distal hereditary motor, autosomal dominant | 12.35 | AD | HMND12 614751 | 2p11.2 | REEP1 609139 | Receptor expression-enhancing protein 1 | Beetz et al (2012) | Allelic to HMNR6 (group 12) and SPG31 (group 15) |
| Neuronopathy, distal hereditary motor, autosomal dominant 13 | 12.36 | AD | HMND13 619112 | 11q12.3 | BSCL2 606158 | Seipin | Windpassinger et al (2004) | allelic to SPG17 (group 15) |
| Neuronopathy, distal hereditary motor, autosomal dominant 14 | 12.37 | AD | HMND14 607641 | 2p13.1 | DCTN1 601143 | Dynactin 1 | Puls et al (2003) | allelic to ALS related to DCTN1 (group 12) |
| Dominant distal hereditary motor neuropathy | 12.38 | AD | dHMN | 16q22.1 | AARS1 601065 | Alanyl-tRNA synthetase 1 | Zhao et al (2012) | allelic to CMT2N (group 14) |
| Hereditary motor and sensory neuropathy V | 12.39 | AD | HMSN5 600361 | 4q34.3-q35.2 | ? | ? | Muglia et al (2008) | |
| Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 1 | 12.40 | 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 | 12.41 | 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 | 12.42 | 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 | 12.43 | 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 | 12.44 | 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) |
| Neuronopathy, Distal hereditary motor related to SYT2 | 12.45 | AD | | 1q32.1 | SYT2 600104 | Synaptotagmin 2 | Montes-Chinea et al (2018) | Allelic to CMS7A, CMS7B (group 11) |
| Neuronopathy, Distal hereditary motor related to GBF1 | 12.46 | AD | | 10q24.32 | GBF1 603698 | Golgi-specific brefeldin-A resistance factor 1 | Mendoza-Ferreira et al (2020) | Allelic to CMT2GG (group 14) |
| Spinal muscular atrophy X linked | | | | | | | | |
| Spinal and bulbar muscular atrophy, X-linked, 1 (Kennedy disease) | 12.47 | XLR | SMAX1 313200 | Xq12 | AR 313700 | Androgen receptor | Fishbeck et al. (1986) La Spada et al. (1991) | |
| Spinal muscular atrophy, distal, X-linked 2 | 12.48 | XLR | SMAX2 301830 | Xp11.3 | UBA1 314370 | Ubiquitin-like modifier-activating enzyme 1 | Ramsler et al (2013) Diamini et al (2013) | |
| Spinal muscular atrophy, distal, X-linked, 3 | 12.49 | XLR | SMAX3 300489 | Xq21.1 | ATP7A 300011 | ATPase, Cu ⁺⁺ transporting, alpha polypeptide | Takata et al (2004) Kennerson et al (2010) | |
| Spinal muscular atrophy with respiratory distress (SMARD) | 12.50 | XLR | | Xq12 | LASIL 300964 | Las1-like ribosome biogenesis factor | Stembalska et al (2022) | |
| Amyotrophic lateral sclerosis (ALS) | | | | | | | | |
| Amyotrophic lateral sclerosis 1 (dominant) | 12.51 | 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) | 12.52 | AR | ALS1 105400 | 21q22.11 | SOD1 147450 | Cu/Zn superoxide dismutase | Andersen et al (1995) | |
| Amyotrophic lateral sclerosis 2, juvenile | 12.53 | 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 IAHS (group 15) |
| Amyotrophic lateral sclerosis 3 | 12.54 | AD | ALS3 606640 | 18q21 | ? | ? | Hand et al (2002) | |
| Amyotrophic lateral sclerosis 4, juvenile | 12.55 | 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) |

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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 5, juvenile | 12.56 | AR | ALS5 602099 | 15q21.1 | <i>SPG11</i> 610844 | Spatacin | Hentati et al (1998) Orlacchio et al (2010) | allelic to CMT2X (group 14) |
| Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia | 12.57 | AD | ALS6 608030 | 16p11.2 | <i>FUS</i> 137070 | Fused in sarcoma | Sapp et al (2003) Abalkhail et al (2003) Kwiatkowski et al (2009) Vance et al (2009) Sapp et al (2003) | |
| Amyotrophic lateral sclerosis 7 | 12.58 | AD | ALS7 608031 | 20p13 | ? | ? | | |
| Amyotrophic lateral sclerosis 8 | 12.59 | AD | ALS8 608627 | 20q13.32 | <i>VAPB</i> 605704 | Vesicle-associated membrane protein-associated protein B and C | Nishimura et al (2004) | allelic to SMAFK (group 12) |
| Amyotrophic lateral sclerosis 9 | 12.60 | AD | ALS9 611895 | 14q11.2 | <i>ANG</i> 105850 | Angiogenin | Greenway et al (2006) Wu et al (2007) | |
| Amyotrophic lateral sclerosis 10, with or without frontotemporal dementia | 12.61 | AD | ALS10 612069 | 1p36.22 | <i>TARDBP</i> 605078 | TAR DNA-binding protein | Sreedharan et al (2008) | |
| Amyotrophic lateral sclerosis 11 | 12.62 | AD | ALS11 612577 | 6q21 | <i>FIG4</i> 609390 | FIG4, <i>S. Cerevisiae</i> , homolog of | Chow et al (2009) | allelic to CMT4J (group 14) |
| Amyotrophic lateral sclerosis 12, with or without frontotemporal dementia | 12.63 | AD, AR | ALS12 613435 | 10p13 | <i>OPTN</i> 602432 | Optineurin | Maruyama et al (2010) | |
| Amyotrophic lateral sclerosis 13, susceptibility to | 12.64 | AD | ALS13 183090 | 12q24.12 | <i>ATXN2</i> 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 | 12.65 | AD | ALS14 613954 | 9p13.3 | <i>VCP</i> 601023 | Valosin-containing protein | Johnson et al (2011) | allelic to Scapuloperoneal myopathy (group 1), IBMPFD1 (groups 4 and 5), CMT2Y (group 14) |
| Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia | 12.66 | XLD | ALS15 300857 | Xp11.21 | <i>UBQLN2</i> 300264 | Ubiquilin 2 | Deng et al (2011) | |
| Amyotrophic lateral sclerosis 16, juvenile | 12.67 | AR | ALS16 614373 | 9p13.3 | <i>SIGMAR1</i> 601978 | Sigma non-opioid intracellular receptor 1 | Al-Saif et al (2011) | allelic to DSMA2 (group 12) donc hereditary distal motor neuropathy |
| Amyotrophic lateral sclerosis 17 | 12.68 | AD | ALS17 614696 | 3p11.2 | <i>CHMP2B</i> 609512 | Charged multivesicular body protein 2B | Parkinson et al (2006) Cox et al (20010) | |
| Amyotrophic lateral sclerosis 18 | 12.69 | AD | ALS18 614808 | 17p13.2 | <i>PFN1</i> 176610 | Profilin 1 | Wu et al (2012) | |
| Amyotrophic lateral sclerosis 19 | 12.70 | AD | ALS19 615515 | 2q34 | <i>ERBB4</i> 600543 | V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 4 | Takahashi et al (2013) | |
| Amyotrophic lateral sclerosis 20 | 12.71 | AD | ALS20 615426 | 12q13.13 | <i>HNRNPA1</i> 164017 | Heterogeneous nuclear ribonucleoprotein A1 | Kim et al (2013) | allelic to IBMPFD3 (group 3) |
| Amyotrophic lateral sclerosis 21 | 12.72 | AD | ALS21 606070 | 5q31.2 | <i>MATR3</i> 164015 | Matrin 3 | Johnson et al (2014) | allelic to VCPDM (group 4) |
| Amyotrophic lateral sclerosis 22, with or without frontotemporal dementia | 12.73 | AD | ALS22 616208 | 2q35 | <i>TUBA4A</i> 191110 | Tubulin, alpha-4A | Smith et al (2014) | |
| Amyotrophic lateral sclerosis 23 | 12.74 | AD | ALS23 617839 | 10q22.3 | <i>ANXA11</i> 602572 | Annexin A11 | Smith et al (2017) | Allelic to IBMWMA (group 12) |
| Amyotrophic lateral sclerosis 24, susceptibility to | 12.75 | | ALS24 617892 | 4q33 | <i>NEK1</i> 604588 | Never in mitosis gene A-related kinase 1 | Brenner et al (2016) Kenna et al (2016) | |
| Amyotrophic lateral sclerosis 25, susceptibility to | 12.76 | AD | ALS25 617921 | 12q13.3 | <i>KIF5A</i> 602821 | Kinesin family member 5A | Nicolas et al. (2018) | Allelic to CMT2 related to KIF5A (group 14) and SPG10 (group 15) |
| Amyotrophic lateral sclerosis 28 | 12.77 | AD | ALS28 620452 | 8q22.3 | <i>LRP12</i> 618299 | Low density lipoprotein receptor-related protein 12 | Kume et al (2023) | Allelic to OPDM1 (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) |
|--|-------------------------|-------------|---|------------|-----------------------------|---|---|---|
| Amyotrophic lateral sclerosis related to NEFH, susceptibility to | 12.78 | AD, AR | ALS1 105400 | 22q12.2 | <i>NEFH</i> 162230 | Neurofilament, heavy polypeptide | Al-Chalabie et al (1999) | allelic to CMT2CC (group 14) |
| Amyotrophic lateral sclerosis related to peripherin, susceptibility to | 12.79 | AD | ALS1 105400 | 12q13.12 | <i>PRPH</i> 170710 | Peripherin | Gros-Louis et al (2004) Leung et al (2004) | |
| Amyotrophic lateral sclerosis related to dynactin 1, susceptibility to | 12.80 | AD | ALS1 105400 | 2p13.1 | <i>DCTN1</i> 601143 | Dynactin 1 | Munch et al (2005) | allelic to HMND14 (group 12) |
| Frontotemporal dementia and/or amyotrophic lateral sclerosis 1 | 12.81 | AD | FTDALS1 105550 | 9p21.2 | <i>C9orf72</i> 614260 | Chromosome 9 open reading frame 72 | Morita et al (2006) DeJesus-Hernandez (2011) | |
| Frontotemporal dementia and/or amyotrophic lateral sclerosis 2 | 12.82 | AD | FTDALS2 615911 | 22q11.23 | <i>CHCHD10</i> 615903 | Coiled-coil-helix-coiled-coil-helix domain containing 10 (M) | Bannwarth et al (2014) | allelic to SMAJ (group 12), IMMD (group 16) |
| Frontotemporal dementia and/or amyotrophic lateral sclerosis 3 | 12.83 | AD | FTDALS3 66437 | 5q35.3 | <i>SQSTM1</i> 601530 | Sequestosome 1 | Fecto et al (2011) | allelic to Welander-like syndrome and DMRV (group 4) |
| Frontotemporal dementia and/or amyotrophic lateral sclerosis 4 | 12.84 | AD | FTDALS4 616439 | 12q14.2 | <i>TBK1</i> 604834 | Tank-binding kinase 1 | Cirulli et al. (2015) Freischmidt et al. (2015) | |
| Amyotrophic lateral sclerosis | 12.85 | AD | | 14q11.2 | <i>LRP10</i> 609921 | Low Density Lipoprotein Receptor-Related Protein 10 | Ni et al (2021) | |
| Amyotrophic lateral sclerosis | 12.86 | AD | | 9q22.31 | <i>SPTLC1</i> 605712 | Serine palmitoyl transferase long-chain base subunit 1 | Mohassel et al (2021) | Allelic to HSAN1A (group 14) |
| Others | | | | | | | | |
| Lethal Congenital Contracture Syndrome 1 | 12.87 | AR | LCCS1 253310 | 9q34.11 | <i>GLE1</i> 603371 | GLE1, RNA export mediator | Makela-Bengs et al (1998) Nousiainen et al. (2008) | |
| Lethal Congenital Contracture Syndrome 2 | 12.88 | AR | LCCS2 607598 | 12q13.2 | <i>ERBB3</i> 190151 | V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 3 | Narkis et al (2007) | |
| Lethal Congenital Contracture Syndrome 3 | 12.89 | AR | LCCS3 611359 | 19p13.3 | <i>PIPSKIC</i> 606102 | Phosphatidylinositol-4-phosphate 5-kinase, type I, gamma | Narkis et al (2007) | |
| Lethal Congenital Contracture Syndrome 4 | 12.90 | AR | LCCS4 614915 | 12q23.2 | <i>MYBPCI</i> 160794 | Myosin-binding protein C, slow type | Markus et al (2012) | Allelic to CMYP16 (group 3), DA1B (group 16) |
| Lethal Congenital Contracture Syndrome 5 | 12.91 | AR | LCCS5 615368 | 19p13.2 | <i>DNM2</i> 602378 | Dynamitin 2 | Koutsopoulos et al (2013) | Allelic to CMD related to <i>DNM2</i> (group 2), CNM1 (group 3), Distal myopathy related to <i>DNM2</i> (group 4), and CMTDIB (group 14) |
| Lethal Congenital Contracture Syndrome 6 | 12.92 | AR | LCCS6 616248 | 14q32.33 | <i>ZBTB42</i> 613915 | Zinc finger and BTB domain-containing protein 42 | Patel et al (2014) | |
| Lethal Congenital Contracture Syndrome 7 | 12.93 | AR | LCCS7 616286 | 17q21.2 | <i>CNTNAP1</i> 602346 | Contactin-associated protein 1 | Laquerriere et al (2014) Lakhani et al (2017) | Allelic to CMT related to <i>CNTNAP1</i> (group 14) |
| Lethal Congenital Contracture Syndrome 8 | 12.94 | AR | LCCS8 616287 | 12q13.12 | <i>ADCY6</i> 600294 | Adenylate cyclase 6 | Laquerriere et al (2014) Agolini et al (2020) | |
| Lethal Congenital Contracture Syndrome 9 | 12.95 | AR | LCCS9 616503 | 6q24.2 | <i>ADGRG6</i> 612243 | Adhesion G protein-coupled receptor G6 | Ravenscroft et al (2015) | |
| Lethal Congenital Contracture Syndrome 10 | 12.96 | AR | LCCS10 617022 | 14q24.3 | <i>NEK9</i> 609798 | Never in mitosis gene A-related kinase 9 | Casey et al (2016) | |
| Lethal Congenital Contracture Syndrome 11 | 12.97 | AR | LCCS11 617194 | 15q21.2 | <i>GLDN</i> 608603 | Gliomedin | Maluenda et al (2016) | |
| Lethal Congenital Contracture Syndrome related to <i>TTN</i> | 12.98 | AR | | 2q31.2 | <i>TTN</i> 188840 | Titin | Chervinski et al (2018) | Allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> , MmD related to <i>TTN</i> and <i>CMYP5</i> (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A) |

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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) |
|---|-------------------------|-------------|---|------------|-----------------------------|--|--|--|
| Spinal muscular atrophy with pontocerebellar hypoplasia, type 1b | 12.99 | AR | PCH1B 614678 | 9p13.2 | EXOSC3 606489 | Exosome component 3 | Rudnik-Schöneborn et al (2013) | |
| Spinal muscular atrophy with or without pontocerebellar hypoplasia | 12.100 | AR | PCH1 607596 | 14q32.2 | VRK1 602168 | Vaccinia related kinase 1 | Renbaum et al (2009) Stoll et al (2016) | Allelic to HMNR10 (group 12), Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis (group 14) |
| Pontocerebellar hypoplasia with spinal muscular atrophy | 12.101 | AR | HSMN6B 616505 | 5q22.1 | SLC25A46 610826 | Solute Carrier Family 25, Member 46 | van Dijk et al (2017) | |
| Brown-Vialetto-van Laere syndrome 1 | 12.102 | 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 | 12.103 | 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) | |
| Late onset spinal muscular atrophy related to <i>HEXB</i> | 12.104 | AR | | 5q13.3 | HEXB 606873 | Hexosaminidase B | Rattay et al (2013) | allelic to Sandhoff disease, adult type (OMIM #268800) |
| Spinal muscular atrophy, related to <i>PRUNE1</i> | 12.105 | AR | | 1q21.3 | PRUNE1 617413 | Prune exopolyphosphatase 1 | Iacomino et al (2017) | |
| Spinal muscular atrophy with pontocerebellar hypoplasia related to <i>KIF26B</i> | 12.106 | AD | | 1q44 | KIF26B 614026 | Kinesin family member 26B | Wojcik et al (2018) | |
| Peripheral neuropathy, myopathy, hoarseness and hearing loss | 12.107 | 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 <i>MAPT</i> | 12.108 | 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 | 12.109 | 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) |
| Inclusion body myopathy and brain white matter abnormalities | 12.110 | AD | IBMWMA 619733 | 10q22.3 | ANXA11 602572 | Annexin A11 | Leoni et al (2021) | allelic to ALS23 (group 12) |
| Mitochondrial complex IV deficiency, nuclear type 11 | 12.111 | AR | MC4DN11 619054 | 1q44 | COX20 614698 | Cytochrome c Oxidase Assembly Factor COX20 | Dong et al (2021) | |
| Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures | 12.112 | AR | CONDSIAS 618170 | 1p34.3 | ADPRHL2 610624 | ADP-Ribosylhydrolase-Like 2 | Danhauser et al (2018) Ghosh et al (2018) | |
| Spinobulbar muscular atrophy (SBMA) | 12.113 | AD | | 9p21.2 | C9orf72 614260 | Chromosome 9 open reading frame 72 | Radziwonik et al (2022) | Allelic to FTDALS1 (group 12) |

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) Street et al (2002, 2003) | allelic to CMTDID, CMT2I, CMT2J, DSS and CHN (group14) |
| 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) |

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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) |
|--|-------------------------|-------------|---|---------------|---|---|---|--|
| Charcot-Marie-Tooth disease, type 1E, with deafness | 14.5 | AD | CMT1E 118300 | 17p12 | <i>PMP22</i> 601097 | Peripheral myelin protein 22 | Kovach et al (1999) Boerkoel et al (2002) | allelic to CMT1A, HNPP and DSS (group14), |
| Hereditary neuropathy with liability to pressure palsies | 14.6 | AD | HNPP 162500 | 17p12 | <i>PMP22</i> 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 | <i>NEFL</i> 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 | <i>PMP2</i> 170715 | Peripheral myelin protein 2 | Hong et al (2016) | |
| Charcot-Marie-Tooth neuropathy | 14.9 | AD | CMT1I 619742 | 12q23.3 | <i>POLR3B</i> 614366 | Polymerase III, RNA, Subunit B | Djordjevic et al (2021) Xue et al (2021) | |
| Charcot-Marie-Tooth with congenital vertical talus | 14.10 | AD | CVT 192950 | 2q31.1 | <i>HOXD10</i> 142984 | Homeobox D10 | Shrimpton et al (2004) | |
| Slowed nerve conduction velocity | 14.11 | AD | SNCV 608236 | 8p23.3 | <i>ARHGEF10</i> 608136 | Rho guanine-nucleotide exchange factor-10 | De Jonghe et al (1999) Verhoeven et al (2003) | |
| Neuropathy, hereditary, with or without age-related macular degeneration | 14.12 | AD | HNARMID 608895 | 14q32.12 | <i>FBLN5</i> 604580 | Fibulin 5 | Auer-Grumbach et al (2011) | |
| Charcot-Marie-Tooth neuropathy | 14.13 | AD | | 6q21.31 | <i>ITPR3</i> 147267 | Inositol 1,4,5-triphosphate receptor, type 3 | Rönkkö et al (2020) | |
| Charcot-Marie-Tooth disease, type 1, related to <i>ATP1A1</i> | 14.14 | AD | | 1p13.1 | <i>ATP1A1</i> 182310 | ATPase, Na ⁺ /K ⁺ transporting, alpha-1 polypeptide | Cinarli Yuksel et al (2023) | |
| Dominant intermediate (CMTDI) | | | | | | | | |
| Charcot-Marie-Tooth disease, dominant intermediate A | 14.15 | AD | CMT2GG 606483 | 10q24.1-q25.1 | <i>GBF1</i> 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 | 14.16 | AD | CMTDIB 606482 | 19p13.2 | <i>DNM2</i> 602378 | Dynamin 2 | Zuchner et al (2005) | allelic to CMD related to <i>DNM2</i> (group 2), <i>CNM1</i> (group 3), distal myopathy related to <i>DNM2</i> (group 4) and <i>LCCS5</i> (group 12) |
| Charcot-Marie-Tooth disease, dominant intermediate C | 14.17 | AD | CMTDIC 608323 | 1p35.1 | <i>YARS</i> 603623 | Tyrosyl-tRNA synthetase | Jordanova et al (2003, 2006) | Allelic to MLASA2 (group 16) |
| Charcot-Marie-Tooth disease, dominant intermediate D | 14.18 | AD | CMTDID 607791 | 1q23.3 | <i>MPZ</i> 159440 | Myelin protein zero | Mastaglia et al (1999) | allelic to CMT1B, CMT2I, CMT2J, DSS and CHN (group14) |
| Charcot-Marie-Tooth disease, dominant intermediate E | 14.19 | AD | CMTDIE 614455 | 14q32.33 | <i>INF2</i> 610982 | Inverted formin 2 | Boyer et al (2011) | |
| Charcot-Marie-Tooth disease, dominant intermediate F | 14.20 | AD | CMTDIF 615185 | 3q28.33 | <i>GNB4</i> 610863 | Guanine nucleotide-binding protein, beta-4 | Soong et al (2013) | |
| Charcot-Marie-Tooth disease, dominant intermediate G | 14.21 | AD | CMTDIG 617882 | 8p21.2 | <i>NEFL</i> 162280 | Neurofilament, light polypeptide 68kDa | Berciano et al (2015) | allelic to CMT1F and CMT2E (group 14) |
| Charcot-Marie Tooth disease, intermediate | 14.22 | AD | | 1p21.2-p13.3 | <i>CFAP276</i> (<i>C1orf194</i>) 618682 | Cilia- and flagella-associated protein 276 | Sun et al (2019) | |
| Charcot-Marie-Tooth disease, recessive intermediate, A | 14.23 | AR | CMTRIA 608340 | 8q21.11 | <i>GDAP1</i> 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 | 14.24 | AR | CMTRIB 613641 | 16q23.1 | <i>KARS</i> 601421 | Lysyl-tRNA synthetase | McLaughlin et al (2010) | |
| Charcot-Marie-Tooth disease, recessive intermediate, C | 14.25 | AR | CMTRIC 615376 | 1p36.31 | <i>PLEKHG5</i> 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 | 14.26 | AR | CMTRID 616039 | 12q24.31 | <i>COX6A1</i> 602072 | Cytochrome c oxidase subunit 6a1 (M) | Tamiya et al (2014) | |

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|--|-------------------------|-------------|---|------------|---|---|--|---|
| Charcot-Marie-Tooth disease, intermediate, related to <i>SARS1</i> | 14.27 | AD | | 1p13.3 | <i>SARS1</i> 607539 | Seryl-tRNA synthetase 1 | He et al (2023) | |
| Autosomal recessive (AR-CMT1 or CMT4) | | | | | | | | |
| Charcot-Marie-Tooth disease, type 4A | 14.28 | AR | CMT4A 214400 | 8q21.11 | <i>GDAP1</i> 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 | 14.29 | AR | CMT4B1 601382 | 11q21 | <i>MTMR2</i> 603557 | Myotubularin-related protein-2 | Bolino et al (1996, 2000) Previtali et al (2003) | |
| Charcot-Marie-Tooth disease, type 4B2 | 14.30 | AR | CMT4B2 604563 | 11p15.4 | <i>SBF2</i> 607697 | SET-binding factor 2 | Azzedine et al (2003) | |
| Charcot-Marie-Tooth disease, type 4B3 | 14.31 | AR | CMT4B3 615284 | 22q13.33 | <i>SBF1</i> 603560 | SET-binding factor 1 | Senderek et al (2004) Nakhro et al (2013) | |
| Charcot-Marie-Tooth disease, type 4C | 14.32 | AR | CMT4C 601596 | 5q32 | <i>SH3TC2</i> 608206 | SH3 domain and tetratricopeptide repeats domain 2 | LeGuern et al (1996) Senderek et al (2003) | |
| Charcot-Marie-Tooth disease, type 4D | 14.33 | AR | CMT4D 601455 | 8q24.22 | <i>NDRG1</i> 605262 | N-myc downstream regulated gene 1 | Kalaydjieva et al (1996, 2000) Hunter et al (2003) | |
| Neuropathy, congenital hypomyelinating, 1 | 14.34 | AR | CHN1 605253 | 10q21.3 | <i>EGR2</i> 129010 | Early growth response 2 | Warner et al (1998) | allelic to CMT1D and DSS (group 14) |
| Neuropathy, congenital hypomyelinating, 2 | 14.35 | AR | CHN2 605253 | 1q23.3 | <i>MPZ</i> 159440 | Myelin protein zero | Warner et al. (1996) | allelic to CMT1B, CMTDD, CMT2I, CMT2J, DSS (group 14) |
| Charcot-Marie-Tooth disease, type 4F | 14.36 | AR | CMT4F 614895 | 19q13.2 | <i>PRX</i> 605725 | Periaxin | Delague et al (2000) Guilbot et al (2001) | allelic to DSS (group 14) |
| Neuropathy, hereditary motor and sensory, Russe type | 14.37 | AR | HMSNR 605285 | 10q22.1 | <i>HK1</i> 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 | 14.38 | AR | CMT4H 609311 | 12p11.21 | <i>FGD4</i> 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 | 14.39 | AR | CMT4J 611228 | 6q21 | <i>FIG4</i> 609390 | FIG4, S. Cerevisiae, homolog of | Chow et al (2007) | allelic to ALS11 (group 12) |
| Charcot-Marie-Tooth disease, type 4K | 14.40 | AR | CMT4K 616684 | 9q34.2 | <i>SURF1</i> 185620 | Surfeit 1 (M) | Echaniz-Laguna et al (2013) | |
| Charcot-Marie-Tooth disease | 14.41 | AR | | 14q32 | <i>AHNAK2</i> 608570 | Ahnak nucleoprotein 2 | Tey et al (2019) | |
| Charcot-Marie-Tooth disease related to <i>CNTNAP1</i> | 14.42 | AR | | 17q21.2 | <i>CNTNAP1</i> 602346 | Contactin-associated protein 1 | Freed et al (2019) | Allelic to LCCS7 (group 12) |
| X-linked CMT1 | | | | | | | | |
| Charcot-Marie-Tooth neuropathy, X-linked dominant, 1 | 14.43 | XLD | CMTX1 302800 | Xq13.1 | <i>GJBI</i> 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 | 14.44 | XLR | CMTX2 302801 | Xp22.2 | ? | ? | Ionasecu et al (1992) | |
| Charcot-Marie-Tooth neuropathy, X-linked recessive, 3 | 14.45 | XLR | CMTX3 302802 | Xq26 | 78kb Chr8 8q24.3 insertion | 78kb 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) | 14.46 | XLR | COWCK 310490 | Xq26.1 | <i>AIFM1</i> 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 | 14.47 | XLR | CMTX5 311070 | Xq22.3 | <i>PRPS1</i> 311850 | Phosphoribosyl pyrophosphate synthetase 1 | Kim et al (2007) | |
| Charcot-Marie-Tooth disease, X-linked dominant, 6 | 14.48 | XLD | CMTX6 300905 | Xp22.11 | <i>PDK3</i> 300906 | Pyruvate dehydrogenase kinase, isoenzyme 3 | Kennerson et al (2013) | |
| Charcot-Marie-Tooth disease, X-linked recessive related to <i>DRP2</i> | 14.49 | XLR | | Xq22.1 | <i>DRP2</i> 300052 | Dystrophin-related protein 2 | Brennan et al (2015) Tahmasebi-Birgani et al (2019) | |

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|--|-------------------------|-----------------|---|------------------|---|--|--|--|
| Déjerine-Sottas syndrome (DSS or CMT3) | | | | | | | | |
| Déjerine-Sottas hypertrophic neuropathy, dominant | 14.50 | 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 | 14.51 | 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 | 14.52 | 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 | 14.53 | 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 | 14.54 | AD | CMT2A1 118210 | 1p36.22 | KIF1B 605995 | Kinesin family member 1B | Zhao et al (2001) | |
| Charcot-Marie-Tooth disease, axonal, type 2A2A | 14.55 | AD | CMT2A2A 609260 | 1p36.22 | MFN2 608507 | Mitofusin 2 | Ben Othmane et al (1993) Züchner et al (2004) | allelic to CMT2A2B and HMSN6A (group14) |
| Charcot-Marie-Tooth disease, type 2B | 14.56 | AD | CMT2B 600882 | 3q21.3 | RAB7 602298 | RAS-associated protein RAB7 | Kwon et al (1995) Pericak-Vance et al (1997) Kok et al (2003) Klein et al (2003) | |
| Hereditary motor and sensory neuropathy, type IIC | 14.57 | AD | HMSN2C 606071 | 12q24.11 | TRPV4 605427 | Transient receptor potential cation channel, subfamily V, member 4 | McEntagart et al (2005) Auer-Grumbach et al (2010) Deng et al (2010) Landouere et al (2010) | allelic to HMND8 and SPSMA (group 12) |
| Charcot-Marie-Tooth disease, type 2D | 14.58 | AD | CMT2D 601472 | 7p14.3 | GARS1 600287 | Glycyl-tRNA synthetase | Ionasescu et al (1996) Antonellis et al (2003) | allelic to HMND5 (group12) |
| Charcot-Marie-Tooth disease, type 2E | 14.59 | 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 | 14.60 | 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 HMND3 (group 14) |
| Charcot-Marie-Tooth disease, type 2H | 14.61 | AD | CMT2H 607731 | 8q13-q23 | ? | ? | Barhoumi et al (2001) | maybe allelic to CMT4A (group14) |
| Charcot-Marie-Tooth disease, type 2I | 14.62 | 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 | 14.63 | AD | CMT2J 607736 | 1q23.3 | MPZ 159440 | Myelin protein zero | De Jonghe et al (1999) Chapou et al (1999) | allelic to CMT1B, CMTDID, CMT2I, DSS, CHN (group 14) |
| Charcot-Marie-Tooth disease, type 2K | 14.64 | 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 | 14.65 | 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 HMND2 (group 12) |
| Charcot-Marie-Tooth disease, type 2N | 14.66 | 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 | 14.67 | 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 | 14.68 | AD | CMT2P 614436 | 9q33.3-q34.1 | LRSA1 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 | 14.69 | AD | CMT2Q 615025 | 10p14 | DHTKDI 614984 | Dehydrogenase E1 and transketolase domain containing 1 | Xu et al (2012) | |
| Charcot-Marie-Tooth disease, type 2U | 14.70 | AD | CMT2U 616280 | 12q13.3 | MARS1 156560 | Methionyl-tRNA synthetase | Gonzalez et al (2013) | |
| Charcot-Marie-Tooth disease, type 2V | 14.71 | AD | CMT2V 616491 | 17q21.2 | NAGLU 609701 | N-acetyl-alpha-glucosaminidase | Tetreault et al (2015) | |

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|--|-------------------------|-------------|---|------------|---|---|--|--|
| Charcot-Marie-Tooth disease, type 2W | 14.72 | AD | CMT2W 616625 | 5q31.3 | <i>HARS</i> 142810 | Histidyl-tRNA synthetase | Vester et al (2013) Safka-Brozkova et al (2015) | |
| Charcot-Marie-Tooth disease, type 2Y | 14.73 | AD | CMT2Y 616687 | 9p13.3 | <i>VCP</i> 601023 | Valosin-containing protein | Gonsalez et al (2014) | allelic to Scapuloperoneal myopathy (group 1), IBMPFD1 (groups 4 and 5), ALS14 (group 12) |
| Charcot-Marie-Tooth disease, type 2Z | 14.74 | AD | CMT2Z 616688 | 22q12.2 | <i>MORC2</i> 616661 | MORC family CW-type zinc finger 2 | Albulym et al (2015) Sevilla et al (2016) | |
| Charcot-Marie-Tooth disease, axonal, type 2CC | 14.75 | AD | CMT2CC 616924 | 22q12.2 | <i>NEFH</i> 162230 | Neurofilament Protein, Heavy Polypeptide | Rebello et al (2016) | allelic to ALS related to NEFH (group 12) |
| Charcot-Marie-Tooth disease, axonal, type 2DD | 14.76 | AD | CMT2DD 618036 | 1p13.1 | <i>ATPIA1</i> 182310 | ATPase, Na ⁺ /K ⁺ transporting, alpha-1 polypeptide | Lassuthova et al (2018) | Allelic to CMT related to ATP1A1 (group 14) |
| Charcot-Marie-Tooth disease, axonal, type 2FF | 14.77 | AD | CMT2FF 619519 | 1q23.2 | <i>CADM3</i> 609743 | Cell adhesion molecule 3 | Rebello et al (2021) | |
| Hereditary motor and sensory neuropathy, Okinawa type | 14.78 | AD | HMSNO 604484 | 3q12.2 | <i>TFG</i> 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 | 14.79 | AD | HMSN6A 601152 | 1p36.22 | <i>MFN2</i> 608507 | Mitofusin 2 | Zuchner et al. (2006) | Allelic to CMT2A2A and CMT2A2B (group 14) |
| CMT2 related to <i>KIF5A</i> | 14.80 | AD | | 12q13.3 | <i>KIF5A</i> 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 | 14.81 | AD | CMT2 | 1q13.5 | <i>DGAT2</i> 606983 | Diacylglycerol O-acyltransferase 2 | Hong et al (2016) | |
| Charcot-Marie-Tooth disease, axonal, related to <i>BAG3</i> | 14.82 | AD | | 10q26.11 | <i>BAG3</i> 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 <i>JAG1</i> | 14.83 | AD | CMT2 | 20p12.2 | <i>JAG1</i> 601920 | Jagged 1 | Sullivan et al (2020) | |
| Charcot-Marie-Tooth disease, axonal, related to <i>NOTCH2NLC</i> | 14.84 | AD | CMT2 | 1q21.2 | <i>NOTCH2NLC</i> (<i>NBPF19</i>) 618025 | Notch2 N-terminal-like protein | Liao et al (2021) | Allelic to OPDM3 (group 5) and Neuronal intranuclear inclusion disease (group 14) |
| Charcot-Marie-Tooth disease, axonal, related to <i>CRYAB</i> | 14.85 | AD | CMT2 | 11q23.1 | <i>CRYAB</i> 123590 | Alpha-B crystallin | Cortese et al (2023) | Allelic to MFM2 (group 5) and CMD1II (group 10A) |
| Charcot-Marie-Tooth disease, axonal, related to <i>DHX9</i> | 14.86 | AD | CMT2 | 1q25.3 | <i>DHX9</i> 603115 | DexH-box helicase 9 | Calame et al (2023) | |
| Autosomal recessive CMT2 | | | | | | | | |
| Charcot-Marie-Tooth disease, axonal, type 2A2B | 14.87 | AR | CMT2A2B 617087 | 1p36.22 | <i>MFN2</i> 608507 | Mitofusin 2 | Polke et al (2011) | allelic to CMT2A2A and HMSN6A (group14) |
| Charcot-Marie-Tooth disease, axonal, type 2B1 | 14.88 | AR | CMT2B1 605588 | 1q22 | <i>LMNA</i> 150330 | Lamin A/C | Bouhouch et al (1999) De Sandre et al (2002) Worman and Bonne (2007) | Allelic to EDMD2, EDMD3 (group 1), MDCL (group 2), CMD1A (group 10A) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermatopathy #275210, MADA #248370] Allelic to AOA4 (group 14) |
| Charcot-Marie-Tooth disease, axonal, type 2B2 | 14.89 | AR | CMT2B2 605589 | 19q13.33 | <i>PNKP</i> 605610 | Polynucleotide kinase 3-prime phosphatase | Leal et al (2009) Leal et al (2018) | |
| Charcot-Marie-Tooth disease, axonal, type 2EE | 14.90 | AR | CMT2EE 618400 | 2p23.3 | <i>MPV17</i> 137960 | MPV17, mouse, homolog of (M) | Choi et al (2015) Baumann et al (2019) | |
| Charcot-Marie-Tooth disease, axonal, related to <i>DNAJB2</i> | 14.91 | AR | | 2q35 | <i>DNAJB2</i> 604139 | DnaJ/Hsp40 homolog, subfamily B, member 2 | Gess et al (2014) | allelic to DSMA5 (group 12) |
| Neuromyotonia and axonal neuropathy, autosomal recessive | 14.92 | AR | NMAN 137200 | 5q23.3 | <i>HINT1</i> 601314 | Histidine triad nucleotide binding protein 1 | Zimon et al (2012) | Allelic to Distal hereditary motor neuronopathy (group 12) |
| Charcot-Marie-Tooth disease, type 2R | 14.93 | AR | CMT2R 615490 | 4q31.3 | <i>TRIM2</i> 614141 | Tripartite motif-containing protein 2 | Ylikallio et al. (2013) Pehlivan et al (2015) | |
| Charcot-Marie-Tooth disease, type 2S | 14.94 | AR | CMT2S 616155 | 11q13.3 | <i>IGHMBP2</i> 600502 | Immunoglobulin mu-binding protein 2 | Cottenie et al (2014) Schottmann et al (2015) | allelic to DSMA1 (group 12) |

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|--|-------------------------|-------------|---|------------|-----------------------------|--|---------------------------|---|
| Charcot-Marie-Tooth disease, type 2T | 14.95 | AR | CMT2T 617017 | 3q25.2 | <i>MME</i> 120520 | Membrane metalloendopeptidase | Higuchi et al (2016) | allelic to SCA43 (group 15) |
| Charcot-Marie-Tooth disease, type 2X | 14.96 | AR | CMT2X 616668 | 15q21.1 | <i>SPG11</i> 610844 | SPG11 vesicle trafficking associated Spatacsin | Montecchiani et al (2015) | allelic to ALS5 (group 12) |
| Early-onset axonal Charcot-Marie-Tooth with ataxia | 14.97 | AR | AOA4 616267 | 19q13.33 | <i>PNKP</i> 605610 | Polynucleotide kinase 3'-phosphatase | Pedroso et al (2015) | allelic to CMT2B2 (group 14) |
| Charcot-Marie-Tooth disease, axonal | 14.98 | AR | | 10q22.1 | <i>SGPL1</i> 603729 | Sphingosine-1 phosphate lyase 1 | Atkinson et al (2017) | allelic to Nephrotic Syndrome 14 (# 617575) |
| Charcot-Marie-Tooth disease, axonal; related to <i>SCO2</i> | 14.99 | AR | | 22q13.33 | <i>SCO2</i> 604272 | Cytochrome c oxidase assembly protein 2 | Rebelo et al (2018) | allelic to MC4DN2 (CEMCOX1) (group 10A) |
| Charcot-Marie-Tooth disease, axonal; related to <i>SACS</i> | 14.100 | AR | | 13q12.12 | <i>SACS</i> 604490 | Sacsin | Souza et al (2018) | allelic to SACS (group 13) and SPAX6 (group 15) |
| Sorbitol dehydrogenase deficiency with peripheral neuropathy (CMT2 AR) | 14.101 | AR | SORDD 618912 | 15q21.1 | <i>SORD</i> 182500 | Sorbitol Dehydrogenase | Cortese et al (2020) | Allelic with HMNR8 (group 15) |
| Charcot-Marie-Tooth disease, axonal | 14.102 | AR | | 21q22.3 | <i>MCM3AP</i> 603294 | Minichromosome maintenance 3-associated protein | Sedghi et al (2019) | allelic to PNRIID (group 12) |
| Charcot-Marie-Tooth disease, axonal | 14.103 | AR | | 12q13.3 | <i>B4GALNT1</i> 601823 | Beta-1,4-N-Acetylgalactosaminyl transferase 1 | Hong et al (2021) | allelic to SPG26 (group 15) |
| Charcot-Marie-Tooth disease, axonal | 14.104 | AR | | 19p13.11 | <i>MYO9B</i> 602129 | Myosin IXB | Cipriani et al (2022) | |
| Progressive neuropathy | 14.105 | AR | | 9q21.2 | <i>PSAT1</i> 610936 | Phosphoserine Aminotransferase 1 | Debs et al (2021) | |

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 | 14.105 | AD | HSAN1A 162400 | 9q22.31 | <i>SPTLC1</i> 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 | 14.106 | AD | HSAN1B 608088 | 3p24-p22 | ? | ? | Kok et al (2004) | |
| Hereditary sensory and autonomic neuropathy, type IC | 14.107 | AD | HSAN1C 613640 | 14q24.3 | <i>SPTLC2</i> 605713 | Serine palmitoyl transferase long-chain base subunit 2 | Rotthier et al (2010) | |
| Hereditary sensory and autonomic neuropathy, type IIA | 14.108 | AR | HSAN2A 201300 | 12p.13.33 | <i>WNK1</i> 605232 | Protein kinase, lysine deficient 1 | Lafreniere et al (2004) Shekarabi et al (2008) | |
| Hereditary sensory and autonomic neuropath, type IIB | 14.109 | AR | HSAN2B 613115 | 5p15.1 | <i>RETREG1</i> (<i>FAM134B</i>) 613114 | Family with sequence similarity 134 member B | Kurth et al (2009) | |
| Hereditary sensory and autonomic neuropathy, type IID | 14.110 | AR | CIP 24300 | 2q24.3 | <i>SCN9A</i> 603415 | Sodium channel, voltage-gated alpha subunit | Yuan et al (2013) | allelic to SFNP (group 14) |
| Hereditary sensory and autonomic neuropathy type III | 14.111 | AR | HSAN3 223900 | 9q31.3 | <i>ELP1</i> 603722 | Inhibitor of kappa light polypeptide gene enhancer 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 | 14.112 | AR | CIPA 256800 | 1q23.1 | <i>NTRK1</i> 191315 | Neurotrophic tyrosine kinase, receptor, type 1 | Indo et al (1996) | |
| Hereditary sensory and autonomic neuropathy type V | 14.113 | AR | HSAN5 608654 | 1p13.1 | <i>NGF</i> 162030 | Nerve growth factor (beta polypeptide) | Einarsdottir et al (2004) | |
| Hereditary sensory and autonomic neuropathy type VI | 14.114 | AR | HSAN6 614653 | 6p12.1 | <i>DST</i> 113810 | Dystonin | Edvardson et al (2012) | |
| Neuropathy, hereditary sensory and autonomic, type VII | 14.115 | AD | HSAN7 615548 | 3p22.2 | <i>SCN11A</i> 604385 | Sodium channel, voltage-gated alpha subunit | Leipold et al (2013) | allelic to FEPS3 (group 14) |

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|---|-------------------------|-------------|---|------------|---|---|--|---|
| Hereditary sensory and autonomic neuropathy type VIII | 14.116 | AR | HSAN8 616488 | 9q34.12 | <i>PRDM12</i> 616458 | PR Domain-containing protein 12 | Chen et al (2015) | |
| Hereditary sensory neuropathy, type ID | 14.117 | AD | HSN1D 613708 | 14q22.1 | <i>ATL1</i> 606439 | Atlantin GTPase 1 | Guelly et al (2011) | allelic to SPG3A (group 15) |
| Hereditary sensory neuropathy, type IE | 14.118 | AD | HSN1E 614116 | 19p13.2 | <i>DNMT1</i> 126375 | DNA methyltransferase 1 | Klein et al (2011) | |
| Neuropathy, hereditary sensory, type IF | 14.119 | AD | HSN1F 615632 | 11q13.1 | <i>ATL3</i> 609369 | Atlantin GTPase 3 | Kornak et al (2014) | |
| Hereditary sensory neuropathy, type IIC | 14.120 | AR | HSN2C 614213 | 2q37.3 | <i>KIF1A</i> 601255 | Kinesin family member 1A | Riviere et al (2011) | allelic to SPG30 (group 15) |
| Ataxia, posterior column, with retinitis pigmentosa (PCARP) | 14.121 | AR | AXPC1 609033 | 1q32.3 | <i>FLVCR1</i> 609144 | Feline leukemia subgroup C receptor 1 | Rajadhyaksha et al (2010) | |
| Absence of pain, Congenital | 14.122 | AR | | 22q11.21 | <i>CLTCL1</i> 601273 | Clathrin, heavy polypeptide-like 1 | Nahorski et al (2015) | |
| Marsili syndrome (insensitivity to pain, congenital, AD) | 14.123 | AD | MARSIS 147430 | 14q11.2 | <i>ZFX2</i> 617828 | Zinc finger homeobox 2 | Habib et al (2018) | |
| Erythromelalgia, Primary | 14.124 | AD | SFNP 133020 | 2q24.3 | <i>SCN9A</i> 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 | 14.125 | AD | FEPS3 615552 | 3p22.2 | <i>SCN11A</i> 604385 | Sodium channel, voltage-gated alpha subunit | Zhang et al (2013) Huang et al (2014) | allelic to HSAN7 (group 14) |
| Polyneuropathy with erythromelalgia | 14.126 | AR | | 1q25.3 | <i>NMNAT2</i> 608701 | Nicotinamide nucleoside adenyltransferase 2 | Huppke et al (2019) | |
| Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome | 14.127 | AR | CANVAS 614575 | 4p14 | <i>RFC1</i> 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) | 14.128 | AR | ACCPN 218000 | 15q14 | <i>SLC12A6</i> (<i>KCC3</i>) 604878 | Solute carrier family 12 (potassium/chloride transporter), member 6 | Casaubon et al (1996) Howard et al (2002) | |
| Peripheral neuropathy and deafness, autosomal dominant | 14.129 | AD | | 1p34.3 | <i>GJB3</i> 603324 | Gap junction protein, beta 3 | Lopez-Bigas et al (2001) | |
| Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy) | 14.130 | AD | HNA 162100 | 17q25.3 | <i>SEPT9</i> 604061 | Septin 9 | Pellegrino et al (1996) Kuhlenbaumer et al (2005) | |
| Giant axonal neuropathy-1 | 14.131 | AR | GAN1 256850 | 16q23.2 | <i>GAN1</i> 605379 | Gigaxonin | Ben Hamida et al (1997) Bomont et al (2000) | |
| Giant axonal neuropathy-2 | 14.132 | AD | GAN2 610100 | 1q23.2 | <i>DCAF8</i> 615820 | DDB1- and CUL4-associated factor 8 | Klein et al (2014) | |
| Congenital cataracts, facial dysmorphism and neuropathy | 14.133 | AR | CCFDN 604168 | 18p23 | <i>CTDPI</i> 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 | 14.134 | AR | | 14q32.2 | <i>VRK1</i> 602168 | Vaccinia related kinase 1 | Gonzaga-Jauregui et al (2013) | Allelic to DSMA and PCH1 (group 12) |
| Neuropathy, hereditary sensory, with spastic paraplegia | 14.135 | AR | 256840 | 5p15.2 | <i>CCT5</i> 610150 | Chaperonin containing T-complex polypeptide1, subunit 5 | Bouhouche et al (2006) | |
| Neuronal intranuclear inclusion diseases | 14.136 | AD | 603472 | 1q21.2 | <i>NOTCH2NLC</i> (<i>NBPF19</i>) 618025 | Notch2 N-terminal-like protein | Tian et al (2019) Sone et al (2019) Ishiura et al (2019) | Allelic to OPDM3 (group5) and CMT2 related to <i>NOTCH2NLC</i> (group 14) |
| Hereditary peripheral neuropathy (CMT ?) | 14.137 | AD | | 17q25.1 | <i>SLC9A3R1</i> (<i>EBP50</i>) 604990 | Solute carrier family 9, member 3, regulator 1 | Song et al (2020) | |

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|--|-------------------------|-------------|---|-----------------|-----------------------------|--|--|--|
| Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities | 14.138 | AR, AD | NEDMILG 619091 | 18q21.31 | <i>NARS1</i> 108410 | Asparaginyl-tRNA Synthetase 1 | Manole et al (2020) | |
| Neurodevelopmental disorder with hypotonia, neuropathy and deafness | 14.139 | AR | NEDHND 617519 | 19q13.2 | <i>SPTBN4</i> 606214 | Spectrin, beta, nonerythrocytic, 4 | Knierim et al (2017) | |
| Group 16. OTHER NEUROMUSCULAR DISORDERS | | | | | | | | |
| Torsion dystonia 1, early onset | 16.1 | AD | DYT1 128100 | 9q34.11 | <i>TOR1A</i> 605204 | Torsin 1A | Ozelius et al (1997) Ikeuchi et al (1999) | |
| Myoclonic dystonia 11 | 16.2 | AD | DYT11 159900 | 7q21.3 | <i>SGCE</i> 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 | <i>ELP1</i> 603722 | Inhibitor of kappa Light polypeptide gene enhancer 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 | <i>TTR</i> 176300 | Transthyretin | Costa et al. (1978) Tawara et al. (1983) Saraiva et al (1995) | |
| Fibrosis of extraocular muscles, congenital, 1 | 16.5 | AD | CFEOM1 135700 | 12q12 | <i>KIF21A</i> 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 | <i>PHOX2A</i> 602753 | Aristaless homeobox, drosophila, homolog 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 | <i>TUBB3</i> 602661 | Tubulin, beta-3 | Doherty et al (1999) Tischfield et al (2010) | |
| Fibrosis of extraocular muscles, congenital, 5 | 16.8 | AR | CFEOM5 66219 | 4q25 | <i>COL25A1</i> 610004 | Collagen, type XXV, alpha-1 | Shinwari et al (2015) | |
| Arthrogryposis, distal, type 1A | 16.9 | AD | DA1A 108120 | 9p13.3 | <i>TPM2</i> 190990 | Tropomyosin-2 (beta) | Sung et al (2003) | Allelic to NEM4 and CMYP23 (group 3), DA2B4 (group 16) |
| Arthrogryposis, distal, type 1B | 16.10 | AD | DA1B 614335 | 12q23.2 | <i>MYBPCI</i> 160794 | Myosin-binding proteinC, slow type | Gurnett et al (2010) | Allelic to CMYP16 (group 3), LCCS4 (group 12) |
| Arthrogryposis, distal, type 2A, Freeman-Sheldon syndrome | 16.11 | AD | DA2A 193700 | 17p13.1 | <i>MYH3</i> 160720 | Myosin heavy chain 3, skeletal muscle, embryonic | Toydemir et al (2006) | Allelic to DA2B (group 16) |
| Arthrogryposis, distal, type 2B1, Sheldon-Hall syndrome | 16.12 | AD | DA2B1 601680 | 11p15.5 | <i>TNNI2</i> 191043 | Troponin I, fast skeletal | Sung et al (2003a) Kimber et al (2006) | |
| Arthrogryposis, distal, type 2B2 | 16.13 | AD | DA2B2 618435 | 11p15.5 | <i>TNNT3</i> 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 | <i>MYH3</i> 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 | <i>TPM2</i> 190990 | Tropomyosin-2 (beta) | Tajsharghi et al (2007) Ochala et al (2007) | Allelic NEM4 and CMYP23 (group 3), DA1A (group 16) |
| Arthrogryposis, distal, type 3 | 16.16 | AD | DA3 114300 | 18p11.22-p11.21 | <i>PIEZO2</i> 613629 | Piezo-type mechanosensitive ion channel component 2 | McMillin et al (2014) | Allelic to DA5 (group 16) |
| Arthrogryposis, distal, type 5 | 16.17 | AD | DA5 108145 | 18p11.22-p11.21 | <i>PIEZO2</i> 613629 | Piezo-type mechanosensitive ion channel component 2 | Coste et al (2013) | Allelic to DA3 (group 16) |
| Arthrogryposis, distal, type 5D | 16.18 | AR | DA5D 615065 | 2q37.1 | <i>ECEL1</i> 605896 | Endothelin-converting enzyme like 1 | McMillin et al (2013) | |
| Arthrogryposis, Distal, type 7 (Trismus-pseudocamptodactyly syndrome) | 16.19 | AD | DA7 158300 | 17p13.1 | <i>MYH8</i> 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, distal type 11 | 16.21 | AD | DA11 620019 | 7q31.2 | <i>MET</i> 164860 | MET protooncogene | Zhou et al (2019) | |

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|--|-------------------------|-------------|---|------------|-----------------------------|---|--|--|
| Arthrogryposis, distal type 12 | 16.22 | AR | DA12 620545 | 11q24.3 | ADAMTS15 607509 | A disintegrin-like and metalloproteinase with thrombospondin type 1 motif 15 | Boschann et al (2022) | |
| Arthrogryposis multiplex congenita with nesprin-1 defect | 16.23 | 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 <i>SYNE1</i> (group 10A) and SCAR8 (group 13) |
| Arthrogryposis and BICD2-related neuromuscular disease | 16.24 | AD | | 9q22.31 | BICD2 609797 | Bicaudal D homolog 2 (Drosophila) | Storbeck et al (2017) | Allelic to SMALED2A and SMALED2B (group 12) |
| Arthrogryposis related to <i>ASCCI</i> | 16.25 | AR | | 10q22.1 | ASCCI 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 | 16.26 | AR | NEDMABA 618622 | 2q21.1 | SMPD4 610457 | Sphingomyelin phosphodiesterase 4, neutral membrane | Magini et al (2019) | |
| Fetal akinesia deformation sequence 1 | 16.27 | 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 | 16.28 | 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 | 16.29 | AR | FADS2 618388 | 11p11.2 | RAPSN 601592 | Receptor-associated protein of the synapse, 43kD (Rapsyn) | Vogt et al (2008) | allelic to CMS11 (group 11) |
| Fetal akinesia deformation sequence 4 | 16.30 | AR | FADS4 618393 | 17p13.2 | NUP88 602552 | Nucleoporin 88kD | Bonnin et al. (2018) | |
| Fetal akinesia deformation sequence related to <i>RYR1</i> | 16.31 | AR | | 19q13.2 | RYR1 180901 | Ryanodine receptor | Romero et al (2003) McKie et al (2014) Alkhunaizi et al (2019) | Allelic to Congenital muscular dystrophy related to RYR1 (group 2), CMYP1A, CMYP1B, CCD, CFTD, CNMDU1, Centronuclear myopathy related to RYR1, DuCD related to RYR1 (group 3), Late onset axial myopathy (group 5), MHS1 (group 8) |
| Fetal akinesia deformation related to <i>AGRN</i> | 16.32 | AR | | 1p36.33 | AGRN 103320 | Agrin | Geremek et al (2020) | Allelic to CMS8 (group 11) |
| Severe foetal hypokinesia related to <i>SCN4A</i> | 16.33 | AR | | 17q23.3 | SCN4A 603967 | Sodium channel, voltage-gated, type IV, alpha subunit | Zaharieva et al (2019) | allelic to CMYP22A, CMYP22B (group 3), HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11) allelic to SANDO (group 13), MTDPS4A, MTDPS4B, PEOB1 (group 16) |
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 | 16.34 | AD, AR | PEOA1 157640 (AD) 258450 (AR) | 15q26.1 | POLG 174763 | Polymerase, DNA, gamma (M) | Van Goethem et al (2001) | |
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 | 16.35 | 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 | 16.36 | AD | PEOA3 609286 | 10q24.31 | TWINK 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 | 16.37 | 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 | 16.38 | AD | PEOA5 613077 | 8q22.3 | RRM2B 604712 | Ribonucleotide reductase, M2 B (M) | Tyynismaa et al (2009) | allelic to MTDP8A (group 16) |

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|--|-------------------------|-------------|---|------------|-----------------------------|---|---------------------------|--|
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6 | 16.39 | AD | PEOA6 615156 | 10q21.3 | <i>DNA2</i> 601810 | DNA replication helicase 2 (M) | Ronchi et al. (2013) | |
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 1 | 16.40 | AR | PEOB1 258450 | 15q26.1 | <i>POLG</i> 174763 | Polymerase, DNA, gamma (M) | Deschauer et al (2007) | allelic to SANDO (group 13), MTDP54A, MTDP54B, PEOA1 (group 16) |
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2 | 16.41 | AR | PEOB2 616479 | 2p25.3 | <i>RNASEH1</i> 604123 | Ribonuclease H1 (M) | Reyes et al (2015) | |
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3 | 16.42 | AR | PEOB3 617069 | 16q21 | <i>TK2</i> 188250 | Thymidine kinase, mitochondrial (M) | Tynnismaa et al (2012) | allelic to MTDP52 (group 16) |
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4 | 16.43 | AR | PEOB4 617070 | 2p13.1 | <i>DGUOK</i> 601465 | Deoxyguanosine kinase (M) | Ronchi et al (2012) | Allelic to MTDP53 (group 16) |
| Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5 | 16.44 | AR | PEOB5 618098 | 17p11.2 | <i>TOP3A</i> 601243 | DNA topoisomerase III | Nicholls et al. (2018) | |
| Mitochondrial DNA depletion syndrome 1 (MNGIE type) | 16.45 | AR | MTDPS1 603041 | 22q13.33 | <i>TYMP</i> 131222 | Thymidine phosphorylase | Nishino et al. (1999) | |
| Mitochondrial DNA depletion syndrome 2 (myopathic type) | 16.46 | AR | MTDPS2 609560 | 16q21 | <i>TK2</i> 188250 | Thymidine kinase, mitochondrial (M) | Saada et al (2001) | allelic to PEOB3 (group 16) |
| Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) | 16.47 | AR | MTDPS3 251880 | 2p13.1 | <i>DGUOK</i> 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) | 16.48 | AR | MTDPS4A 203700 | 15q26.1 | <i>POLG</i> 174763 | Polymerase, DNA, gamma (M) | Naviaux and Nguyen (2004) | Allelic to SANDO (group 13), MTDP54B, PEOA1 and PEOB1 (group 16) |
| Mitochondrial DNA depletion syndrome 4B (MNGIE type) | 16.49 | AR | MTDPS4B 613662 | 15q26.1 | <i>POLG</i> 174763 | Polymerase, DNA, gamma (M) | Van Goethem et al. (2003) | Allelic to SANDO (group 13), MTDP54A, PEOA1 and PEOB1 (group 16) |
| Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) | 16.50 | AR | MTDPS5 612073 | 13q14.2 | <i>SUCLA2</i> 603921 | Succinate-CoA ligase, ADP-forming, beta subunit (M) | Elpeleg et al (2005) | |
| Mitochondrial DNA depletion syndrome 8A (Encephalomyopathic type with renal tubulopathy) | 16.51 | AR | MTDP8A 612075 | 8q22.3 | <i>RRM2B</i> 604712 | Ribonucleotide reductase, M2B (M) | Bourdon et al (2007) | allelic to PEOA5 and MTDP58B (group 16) |
| Mitochondrial DNA depletion syndrome 8B (MNGIE type) | 16.52 | AR | MTDP8B 612075 | 8q22.3 | <i>RRM2B</i> 604712 | Ribonucleotide reductase, M2B (M) | Bourdon et al (2007) | allelic to PEOA5 and MTDP58A (group 16) |
| Mitochondrial DNA depletion syndrome 9 (Encephalomyopathic type with methylmalonic aciduria) | 16.53 | AR | MTDP59 245400 | 2p11.2 | <i>SUCLG1</i> 611224 | Succinate-CoA ligase, alpha subunit (M) | Ostergaard et al. (2007) | |
| Mitochondrial DNA depletion syndrome 11 | 16.54 | AR | MTDP511 615084 | 20p11.23 | <i>MGME1</i> 615076 | Mitochondrial genome maintenance exonuclease 1 (M) | Komblum et al. (2013) | |
| Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) | 16.55 | AD | MTDP512A 617184 | 4q35.1 | <i>SLC25A4</i> 103220 | Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 (M) | Thompson et al. (2016) | Allelic to PEOA2 and MTDP512B (group 16) |

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| Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) | 16.56 | AR | MTDPS12B 615418 | 4q35.1 | <i>SLC25A4</i> 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) | 16.57 | AR | MTDPS13 615471 | 6q16.1-q16.2 | <i>FBXL4</i> 605654 | F-box and leucine-rich repeat protein 4 | Bonnen et al. (2013) Gai et al. (2013) | |
| Mitochondrial DNA depletion syndrome 14 (cardioencephalomyopathic type) | 16.58 | AR | MTDPS14 616896 | 3q29 | <i>OPA1</i> 605290 | OPA1 protein (M) | Spiegel et al. (2016) | Allelic to Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia and neuropathy (group 16) |
| Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression | 16.59 | AR | MECREN 618416 | 19p13.11 | <i>SLC25A42</i> 610823 | Solute carrier family 25, member 42 (M) | Shamseldin et al (2015) | |
| Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia and neuropathy | 16.60 | AD | 125250 | 3q29 | <i>OPA1</i> 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 | 16.61 | AR | MLASA1 600462 | 12q24.33 | <i>PUS1</i> 608109 | Pseudourine synthase 1 (M) | Bykhovskaya et al (2004) Fernandez-Vizarra (2007) Riley et al (2010) | Allelic to CMTDIC (group 14) |
| Myopathy, lactic acidosis, and sideroblastic anemia-2 | 16.62 | AR | MLASA2 613561 | 12p11.21 | <i>YARS2</i> 610957 | Tyrosyl-tRNA synthetase 2 (M) | | Allelic to CMTDIC (group 14) |
| Isolated mitochondrial myopathy | 16.63 | AD | IMMD 616209 | 22q11.23 | <i>CHCHD10</i> 615903 | Coiled-coil-helix-coiled-coil-helix domain containing 10 (M) | Ajrout-Driss et al (2014) | allelic to FTDALS2 and SMAJ (group 12) |
| Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis | 16.64 | AD | POIKTMP 615704 | 11q12.1 | <i>FAM111B</i> 615584 | Family with sequence similarity 111, member B | Mercier et al (2015) | |
| Combined oxidative phosphorylation deficiency 6; | 16.65 | XLR | COXPD6 300816 | Xq26.1 | <i>AIFM1</i> 300169 | Apoptosis-inducing factor, mitochondria-associated, 1 (M) | Morton et al (2017) | Allelic to cerebellar ataxia (group 13) and COCK (group 14) |
| Combined oxidative phosphorylation deficiency 43 | 16.66 | AR | COXPD43 618851 | 17q13.3 | <i>TIMM22</i> 607251 | Translocase of inner mitochondrial membrane 22 (M) | Pacheu-Grau et al (2018) | |
| Combined oxidative phosphorylation deficiency 44 | 16.67 | AR | COXPD44 618855 | 2q33.3 | <i>FASTKD2</i> 612322 | Fast kinase domains 2 (M) | Yoo et al (2017) | |
| Combined oxidative phosphorylation deficiency 48 | 16.68 | AR | COXPD48 619012 | 3q11.2 | <i>NSUN3</i> 617491 | NOP2/SUN RNA Methyltransferase Family Member 3 | Van Haute et al (2016) | |
| Combined oxidative phosphorylation deficiency 50 | 16.69 | AR | COXPD50 619025 | 3p25.1 | <i>MRPS25</i> 611987 | Mitochondrial ribosomal protein S25 (M) | Bugiardini et al (2019) | |
| Mitochondrial complex IV deficiency, nuclear type 18 | 16.70 | AR | MC4DN18 619062 | 16p11.2 | <i>COX6A2</i> 6020009 | Cytochrome c-oxidase, subunit 6A2 (M) | Inoue et al (2019) | |
| Mitochondrial myopathy with severe neurological manifestations | 16.71 | AR | | 8q24.13 | <i>TMEM65</i> 616609 | Transmembrane protein 65 | Nasli et al (2017) | |
| Infantile-onset multisystem disease with progressive muscle weakness | 16.72 | AR | IMNEPD 616263 | 17q23.1 | <i>PTRH2</i> 608625 | Peptidyl-tRNA hydrolase 2 | Hu et al (2014) | |
| Coenzyme Q10 deficiency 1 | 16.74 | AR | COQ10D1 607426 | 4q21.23 | <i>COQ2</i> 609825 | Coenzyme Q2 (M) | Quinzii et al (2006) | |
| Autosomal recessive spinocerebellar ataxia, 9 with ubiquinone deficiency (also listed in group 13) | 16.75 | AR | SCAR9 (COQ10D4) 612016 | 1q42.13 | <i>ADCK3</i> (COQ8A, CABC1) 606980 | Coenzyme Q8A (M) | Lagier-Tourenne et al (2008) Mollet et al (2008) | |
| Coenzyme Q10 deficiency 5 | 16.76 | AR | COQ10D5 614654 | 16q21 | <i>COQ9</i> 612837 | Coenzyme Q9 (M) | Rahman et al (2001) Duncan et al (2009) | |
| Coenzyme Q10 deficiency 6 | 16.77 | AR | COQ10D6 614650 | 14q24.3 | <i>COQ6</i> 614647 | Coenzyme Q6 (M) | Heeringa et al (2011) | |

(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) |
|---|-------------------------|-------------|---|------------|-----------------------------|--|---|--|
| Coenzyme Q10 deficiency 7 | 16.78 | AR | COQ10D7 607426 | 9q34.11 | <i>COQ4</i> 612898 | Coenzyme Q4 (M) | Brea-Calvo et al (2015) | |
| Coenzyme Q10 deficiency 8 | 16.79 | AR | COQ10D8 616733 | 16p12.3 | <i>COQ7</i> 601683 | Coenzyme Q7 (M) | Freyer et al (2015) | Allelic to HMNR9 (group 12) |
| Mitochondrial myopathy, episodic with or without atrophy and reversible leukoencephalopathy | 16.80 | AR | MEOAL 251900 | 19p13.2 | <i>FDX2</i> 614585 | Ferredoxin (M) | Gurgel-Giannetti et al (2019) | |
| Spectraplaklinopathy | 16.81 | AR | | 1p34.3 | <i>MACF1</i> 608271 | Microtubule-actin cross-linking factor 1 | Kang et al (2020) | allelic to LIS9 (#618325) |
| Mitochondrial complex I deficiency, nuclear type 29 | 16.82 | AR | MC1DN29 618250 | 11q14.1 | <i>TMEM126B</i> 615533 | Transmembrane protein 126B (M) | Alston et al (2016) Sanchez-Caballero et al (2016) | |
| Mitochondrial myopathy with lactic acidosis, cognitive impairment and autistic features | 16.83 | XL | | Xp22.11 | <i>APOO</i> 300753 | Apolipoprotein O | Beninca et al (2021) | |
| Segmental amyoplasia with Distal Arthrogyposis | 16.84 | AD | DA1C 619110 | 16p11.2 | <i>MYLPP</i> 617378 | Myosin Light Chain Phosphorylatable Fast Skeletal Muscle Growth Factor | Chong et al (2020) | |
| Myopathy, mitochondrial progressive, with congenital cataract and developmental delay | 16.85 | AR | MPMCD 613076 | 16p13.3 | <i>GFER</i> 600924 | ERV1-Like | Di Fonzo et al (2009) | |
| Mitochondrial DNA depletion syndrome 20 (MNGIE type) | 16.86 | AR | MTDPS20 619780 | 17q12 | <i>LIG3</i> 600940 | Ligase III DNA ATP-Dependent | Bonora et al (2021) | |
| Mitochondrial complex IV deficiency, nuclear type 22 | 16.87 | AR | MC4DN22 619355 | 14q24.2 | <i>COX16</i> 618064 | Cytochrome c Oxidase Assembly Factor 16 | Wintjes et al (2021) | |
| Mitochondrial myopathy related to <i>SLC25A26</i> | 16.88 | AR | | 3p14.1 | <i>SLC25A26</i> 611037 | Solute carrier family 25 (mitochondrial carrier, phosphate carrier), member 26 | Schober et al (2022) | |
| Mitochondrial myopathy and multiple mtDNA deletions | 16.89 | AR | | 9q34.11 | <i>ENDOG</i> 600440 | Endonuclease G, mitochondrial | Nasca et al (2022) | |

GT 2024 NEW REFERENCES

GROUP 1. MUSCULAR DYSTROPHIES

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