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# The 2021 version of the gene table of neuromuscular disorders (nuclear genome)

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## General features

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

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.<sup>2</sup> A given gene may be involved in several different clinical entities (phenotypic heterogeneity such as in *LMNA* defects) and conversely a given clinical entity may be produced by a defect in several possible alternative genes (genotypic heterogeneity such as in CMT). In some diseases both kinds of heterogeneity may occur. As a consequence a gene or a disease may be cited in several places of the table.

## The two versions of the gene table<sup>3</sup>

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

The **full online version** contains the complete data of the 16 groups and the cumulative list of key references since 1991. It is freely accessible at <http://www.musclegenetable.fr>. It is designed to cope with the complexity described above. In this version the data are cross-referenced and linked to *PubMed* and to major databases related to molecular

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<sup>1</sup> 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>.

<sup>2</sup> The assigned item number is provisional and may change in the next annual version.

<sup>3</sup> 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.)

medicine (*Leiden Muscular Dystrophy*, *OMIM*, *NCBI*, *GenAtlas*, *Orphanet*, *GeneCards*). It contains several query tools allowing one to perform a variety of interrogations. This computerized version of the table is now surpassing the printed version which cannot accommodate the ever increasing volume and complexity of data. The **statistics tool** instantly provides the latest list of genes, proteins, phenotypes and cumulative bibliographic key references. Each list can be displayed, printed and exported in Excel format.

### Overview of the new data in the 2021 printed version of the gene table (pages 1008 to 1048 of this issue)

There are 37 new items, marked by background shading. Altogether they comprise **18 additional genes**, **1 duplication** on chromosome 11 (item # 2.31) and **18 additional phenotypic variants** caused by a gene already listed in the 2020 version (see box).

The new key references of the printed version of the table are listed on pages 1047-1048 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 2021 version of the gene table of neuromuscular disorders. *Neuromuscul Disord.* 30 (12), 1008-1048.
- Online version: GeneTable of Neuromuscular Disorders: <http://www.musclegenetable.fr>

### Contact

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

### Acknowledgements

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New in the 2021 printed version of the gene table

#### 18 genes added and 1 duplication:

*ADCY6* (item # 12.83)  
*ADGRG6* (item # 12.84)  
*GBF1* (item # 12.39)  
*GIPC1* (item # 5.20)  
*GLDN* (item # 12.86)  
*ITPR3* (item # 14.12)  
*JAG1* (item # 14.74)  
*MCM3AP* (item # 12.98)  
*MPDUI* (item # 2.36)  
*NEK9* (item # 12.85)  
*NMNAT2* (item # 14.115)  
*POPDC3* (item # 1.52)  
*SLC9A3R1* (item # 14.124)  
*SMPD4* (item # 16.25)  
*SORD* (item # 14.93)  
*SVIL* (item # 5.11)  
*UNC45B* (item # 3.63)  
*ZBTB42* (item # 12.81)  
 11q13.2q14.1 duplication (item # 2.31)

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

*ACTN2* (item # 4.25)  
*AGRN* (item # 16.31)  
*CNTNAP1* (item # 12.82 and 14.35)  
*DNAJB6* (item # 4.23)  
*DNM2* (item # 12.80)  
*HSPB8* (item # 4.26)  
*MACF1* (item # 16.79)  
*MYBPC1* (item # 12.79)  
*MYH7* (item # 3.62)  
*MYOT* (item # 10.102)  
*NEB* (item # 4.12)  
*POMK* (item # 1.61)  
*RYR1* (item # 3.64)  
*SCN11A* (item # 14.114)  
*SCN9A* (item # 14.113)  
*SQSTM1 + TIA1* (item # 4.8) (Digenism)  
*TTN* (item # 12.87)

#### 40 new key references

## Gene table of monogenic neuromuscular disorders (nuclear genome only)

Vol. 30 No. 12, December 2020

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

Shaded background indicates newly added items.

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [M])	Key references	Other allelic disease(s) (group in this table)
<b>GROUP 1. MUSCULAR DYSTROPHIES</b>								
Duchenne muscular dystrophy; Becker muscular dystrophy	1.1	XR	DMD 310200 BMD 300376	Xp21.2-p21.1	<i>DMD</i> 300377	Dystrophin	Monaco et al. (1986) Burghes et al. (1987) Koenig et al. (1987, 1988) Hoffman et al. (1987, 1988)	allelic to CMD3B (group10)
Emery-Dreifuss muscular dystrophy, X-linked, type 1	1.2	XR	EDMD1 310300	Xq28	<i>EMD</i> 300384	Emerin	Hodgson et al. (1986) Romeo et al. (1988) Bione et al. (1994, 1995) Klauck et al. (1995) Nigro et al. (1995)	
Emery-Dreifuss muscular dystrophy 6, X-linked	1.3	XR	EDMD6 300696	Xq26.3	<i>FHL1</i> 300163	Four and a half LIM domain 1	Gueneau et al. (2009)	allelic to XMPMA (group 5), SPM (group 5), RBMX1A/B (group 5)
Emery-Dreifuss muscular dystrophy 2, autosomal dominant	1.4	AD	EDMD2 181350	1q22	<i>LMNA</i> 150330	Lamin A/C	Bonne et al. (1999) Worman and Bonne (2007)	allelic to EDMD3 (group 1), formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370]
Emery-Dreifuss muscular dystrophy 3, autosomal recessive	1.5	AR	EDMD3 616516	1q22	<i>LMNA</i> 150330	Lamin A/C	Raffaele di Barletta et al. (2000) Worman and Bonne (2007)	allelic to EDMD2 (group 1), formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370]
Emery-Dreifuss muscular dystrophy 4, autosomal dominant	1.6	AD	EDMD4 612998	6q25.2	<i>SYNE1</i> 608441	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Zhang et al. (2007)	allelic to DCM related to <i>SYNE1</i> (group 10A), SCAR8 (group 13), AMCM (group 16)
Emery-Dreifuss muscular dystrophy 5, autosomal dominant	1.7	AD	EDMD5 612999	14q23.2	<i>SYNE2</i> 608442	Spectrin repeat containing, nuclear envelope 2 (nesprin-2)	Zhang et al. (2007)	
Emery-Dreifuss muscular dystrophy 7, autosomal dominant	1.8	AD	EDMD7 614302	3p25.1	<i>TMEM43</i> (= <i>LUMA</i> ) 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	<i>TOR1AIP1</i> (= <i>LAP1B</i> ) 614512	Torsin A interacting protein 1 (=Lamin Associated Peptide 1B)	Kayman-Kurecki et al. (2014) Fichtman et al. (2019)	

(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	<i>DUX4*</i> 614982 (*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	<i>SMCHD1*</i> 614982 (*causing inappropriate reactivation of DUX4* 606009)	Structural maintenance of chromosomes flexible hinge domain containing 1	de Greef et al. (2010) Sacconi et al. (2012) Lemmers et al. (2012) Sacconi et al. (2013)	allelic to Bosma Arhinia Microphthalmia Syndrome; BAMS (#603457)
Muscular dystrophy with generalized lipodystrophy	1.12	AD	CGL4 613327	17q21.2	<i>CAVIN1</i> 603198	Caveolae-associated protein 1	Hayashi et al. (2009)	
<b>Limb girdle muscular dystrophies, dominant</b>								
LGMD1 (formerly LGDM1E)	1.13	AD	LGMD1 (LGMD1E) 603511	7q36.3	<i>DNAJB6</i> 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)
LGMD2 (Formerly LGMD1F)	1.14	AD	LGMD2 (LGMD1F) 608423	7q32.1	<i>TNPO3</i> 610032	Transportin 3	Palenzuela et al. (2003) Melià et al. (2013) Torella et al. (2013)	allelic to Congenital Myopathy related to TNPO3 (group 3)
LGMD3 (Formerly LGMD1G)	1.15	AD	LGMD3 (LGMD1G) 609115	4q21.22	<i>HNRNPDL</i> 607137	Heterogeneous nuclear ribonucleoprotein D-like	Starling et al. (2005) Vieira et al. (2014)	
LGMD4	1.16	AD	LGMD4 (LGMD1I) 618129	15q15.1	<i>CAPN3</i> 114240	Calpain-3	Vissing et al. (2016) Martinez-Thompson et al. (2018)	allelic to LGMDR1 (group 1)
LGMD5	1.17	AD	LGMD5 (BTHLM1) 158810	21q22.3	<i>COL6A1</i> 120220	Collagen type VI subunit alpha 1	Jöbsis et al. (1996)	allelic to LGMDR12 (group 1), UCMD1 and BTHLM1 (group 2)
LGMD5	1.18	AD	LGMD5 (BTHLM1) 158810	21q22.3	<i>COL6A2</i> 120240	Collagen type VI subunit alpha 2	Jöbsis et al. (1996)	allelic to LGMDR22 (group 1); UCMD1, BTHLM1 and Congenital myosclerosis (group 2)
LGMD5	1.19	AD	LGMD5 (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)	allelic to LGMDR22 (group 1); UCMD1 and BTHLM1 (group 2)
Myofibrillar myopathy 3 (Formerly LGMD1A)	1.20	AD	MF3 609200 (LGMD1A 159000)	5q31	<i>MYOT</i> 604103	Myotilin	Speer et al. (1992) Hauser et al. (2000)	allelic to Distal myotilinopathy (group 4), MF3 (group 5), spheroid body myopathy (group 5), LVHT/NC (group 10A)
Emery-Dreifuss muscular dystrophy 2 (Formerly LGMD1B)	1.21	AD	EDMD2 181350 (LGMD1B 159001)	1q22	<i>LMNA</i> 150330	Lamin A/C	van der Kooij et al. (1997) Muchir et al. (2000) Worman and Bonne (2007)	allelic to EDMD2, EDMD3 and formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
Rippling muscle disease 2 (Formerly LGMD1C)	1.22	AD	RMD2 606072 (LGMD1C 607801)	3p25.3	<i>CAV3</i> 601253	Caveolin-3	Minetti et al. (1998) McNally et al. (1998)	allelic to MPDT (group 4), Creatin phosphokinase elevated serum (group 5), RMD2 (group 6), CMH1 (group 10A), LQT9 (group 10B)
Myofibrillar myopathy 1 (Formerly LGMD1 related to DES)	1.23	AD	MF1 601419	2q35	<i>DES</i> 125660	Desmin	Messina et al. (1997) Greenberg et al. (2012) Hedberg et al. (2012)	allelic to formerly LGMD2R (group 1), MF1 with or without ARCV7 (group 5), CMD11 (group 10A), ARVC7 (group 10B)
LGMD1H	1.24	AD	LGMD1H 613530	3p25.1-p23	?	?	Bisceglia et al. (2010)	

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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)
<i>Limb girdle muscular dystrophies, recessive</i>								
LGMDR1 (Formerly LGMD2A)	1.25	AR	LGMDR1 (LGMD2A) 253600	15q15.1	<i>CAPN3</i> 114240	Calpain-3	Beckmann et al. (1991) Young et al. (1992) Richard et al. (1995, 1997)	allelic to LGMD4 (group 1)
LGMDR2 (Formerly LGMD2B)	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)
LGMDR3 (Formerly LGMD2D)	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)	
LGMDR4 (Formerly LGMD2E)	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)	
LGMDR5 (Formerly LGMD2C)	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)	
LGMDR6 (Formerly LGMD2F)	1.30	AR	LGMDR6 (LGMD2F) 601287	5q33.3-q33.3	<i>SGCD</i> 601411	Delta-sarcoglycan	Passos-Bueno et al. (1996) Nigro et al. (1996)	allelic to CMD1L (group 10A)
LGMDR7 (Formerly LGMD2G)	1.31	AR	LGMDR7 (LGMD2G) 601954	17q12	<i>TCAP</i> 604488	Titin-cap (telethonin)	Moreira et al. (1997) Moreira et al. (2000)	allelic to CMD related to <i>TCAP</i> (group 2), CMH25 (group 10A), CMD1N (group 10A)
LGMDR8 (Formerly LGMD2H)	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)
LGMDR9 (Formerly LGMD2I)	1.33	AR	LGMDR9 (MDDGC5) 607155	19q13.32	<i>FKRP</i> 606596	Fukutin related protein	Driss et al. (2000) Brockington et al. (2001a)	allelic to MDDGB5 (group 2), MDDGA5/WWS (group 2), MDDGA5/MEB (group 2)
LGMDR10 (Formerly LGMD2J)	1.34	AR	LGMDR10 (LGMD2J) 608807	2q31.2	<i>TTN</i> 188840	Titin	Hackman et al. (2003)	allelic to CNM related to <i>TTN</i> (group 3), MmD related to <i>TTN</i> (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10), CMD1G (group 10), LCCS related to <i>TTN</i> (group 12)
LGMDR11 (Formerly LGMD2K)	1.35	AR	LGMDR11 (MDDGC1) 609308	9q34.13	<i>POMT1</i> 607423	Protein O-mannosyltransferase 1	Balci et al. (2005) D'Amico et al. (2006)	allelic to WWS/MDDGA1 (group 2) and MDDGB1 (group 2)
LGMDR12 (Formerly LGMD2L)	1.36	AR	LGMDR12 (LGMD2L) 611307	11p14.3	<i>ANO5</i> ( <i>TMEM16E</i> ) 608662	Anoctamin 5	Jarry et al. (2007) Bolduc et al. (2008, 2010) Hicks et al. (2011)	allelic to MMD3 (group 4)
LGMDR13 (Formerly LGMD2M)	1.37	AR	LGMDR13 (MDDGC4) 611588	9q31.2	<i>FKTN</i> 607440	Fukutin	Murakami et al. (2006) Godfrey et al. (2006)	allelic to MDDGA4/FCMD (group 2), MDDGB4/WWS (group 2), CMD1X (group 10A)
LGMDR14 (Formerly LGMD2N)	1.38	AR	LGMDR14 (MDDGC2) 613158	14q24.3	<i>POMT2</i> 607439	Protein O-mannosyl transferase 2	Biancheri et al. (2007)	allelic to MDDGA2/MDDGB2/WWS/MEB (group 2)
LGMDR15 (Formerly LGMD2O)	1.39	AR	LGMDR15 (MDDGC3) 613157	1p34.1	<i>POMGNT1</i> 606822	Protein O-linked mannose beta1,2-N-acetylglucosaminyl transferase 1	Godfrey et al. (2007) Clement et al. (2008) Raducu et al. (2012)	allelic to MDDGA3/MDDGB3/WWS (group 2) and MDDGA3/MEB (group 2)
LGMDR16 (Formerly LGMD2P)	1.40	AR	LGMDR16 (MDDGC9) 613818	3p21.31	<i>DAG1</i> 128239	Dystrophin-associated glycoprotein 1 (alpha-dystroglycan)	Hara et al. (2011)	allelic to MDDGA9 (group 2)
LGMDR17 (Formerly LGMD2Q)	1.41	AR	LGMDR17 (LGMD2Q) 613723	8q24.3	<i>PLEC</i> 601282	Plectin	Gundesli et al. (2010)	allelic to LGMD with Ophthalmoplegia (group 1), EBSMD (group 5), and Myasthenic syndrome related to <i>PLEC</i> (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)
LGMDR18 (Formerly LGMD2S)	1.42	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)
LGMDR19 (Formerly LGMD2T)	1.43	AR	LGMDR19 (MDDGC14) 615352	3p21.31	<i>GMPFB</i> 615320	GDP-mannose pyrophosphorylase B	Carss et al. (2013) Cabrera-Serrano et al. (2015)	allelic to MEB/MDDGA14, MDDGB14 (group 2) and congenital myasthenic syndrome (group 11)
LGMDR20 (Formerly LGMD2U)	1.44	AR	LGMDR20 (MDDGC7) 616052	7p21.2-p21.1	<i>ISPD</i> 614631	Isoprenoid synthase domain containing protein	Tasca et al. (2013)	allelic to WWS/ MDDGA7 (group 2)
LGMDR21 (Formerly LGMD2Z)	1.45	AR	LGMDR21 (LGMD2Z) 617232	3q13.33	<i>POGLUT1</i> 615618	O-Glucosyltransferase 1	Servian-Morilla et al. (2016)	
LGMDR22 (Bethlem myopathy 1)	1.46	AR	LGMDR22 (UCMD1) 254090	21q22.3	<i>COL6A1</i> 120220	Collagen type VI subunit alpha 1	Pan et al. (2003) Giusti et al. (2005)	allelic to LGMD5 (group 1), UCMD1 and BTHLM1 (group 2)
LGMDR22 (Bethlem myopathy 1)	1.47	AR	LGMDR22 (UCMD1) 254090	21q22.3	<i>COL6A2</i> 120240	Collagen type VI subunit alpha 2	Jokela et al. (2019)	allelic to LGMD5 (group 1), UCMD1, BTHLM1 and congenital myosclerosis (group 2)
LGMDR22 (Bethlem myopathy 1)	1.48	AR	LGMDR22 (UCMD1) 254090	2q37.3	<i>COL6A3</i> 120250	Collagen type VI subunit alpha 3	Demir et al. (2002)	allelic to LGMD5 (group 1); UCMD1 and BTHLM1 (group 2)
LGMDR23	1.49	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)
LGMDR24	1.50	AR	LGMDR24 (MDDGC8) 618135	3p22.1	<i>POMGNT2</i> 614828	Protein O-mannose beta-1,4-N-acetylglucosaminyl transferase 2	Endo et al. (2015)	allelic to MDDGA8 (group 2)
LGMDR25 (Formerly LGMD2X)	1.51	AR	LGMDR25 (LGMD2X) 616812	6q21	<i>BVES</i> (= <i>POPDC1</i> ) 604577	Blood vessel epicardial substance	Schindler et al. (2016)	
LGMDR26	1.52	AR	LGMDR26 618848	6q21	<i>POPDC3</i> 605824	Popeye domain-containing protein 3	Vissing et al. (2019)	
Myofibrillar myopathy 1 (Formerly LGMD2R)	1.53	AR	MFM1 601419 (LGMD2R 615325)	2q35	<i>DES</i> 125660	Desmin	Cetin et al. (2013)	allelic to formerly LGMD1 related to <i>DES</i> (group 1), MFM1 with or without ARCV7 (group 5), CMD11 (group 10A) and ARVC7 (group 10B)
Pompe disease (Formerly LGMD2V)	1.54	AR	GSD2 232300	17q25.3	<i>GAA</i> 606800	Glucosidase alpha, acid	Preisler et al. (2013)	allelic to GSD2 (groups 9 and 10A)
Muscular dystrophy with cardiomyopathy and triangular tongue (Formerly LGMD2W)	1.55	AR	MDRCMTT (LGMD2W) 616827	2q14.3	<i>LIMS2</i> (= <i>PINCH2</i> ) 607908	LIM and senescent cell antigen-like domains 2	Chardon et al. (2015)	
Muscle dystrophy with glycosylation defect, type I	1.56	AR	MDDGC15 (CDG10) 612937	1q22	<i>DPM3</i> 605951	Dolichyl-phosphate mannosyltransferase polypeptide 3	Lefebvre et al. (2009)	
Scapuloperoneal muscular dystrophy and dropped head syndrome	1.57	AR	600416	9p13.3	<i>VCP</i> 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	1.58	AR		8q24.3	<i>PLEC</i> 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 PYROXD1	1.59	AR		12p12.1	<i>PYROXD1</i> 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 KBTBD13	1.60	AD		15q22.31	<i>KBTBD13</i> 613727	Kelch repeat and BTB/POZ domain containing protein 13	Garibaldi et al. (2018)	allelic to NEM6 (group 3)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C12	1.61	AR	MDDGC12 616094	8p11.21	<i>POMK</i> 615247	Protein-O-mannose kinase	Di Costanzo et al. (2014)	Allelic to MDDGA12 (group 2)

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<b>GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES</b>								
Congenital muscular dystrophy with merosin deficiency	2.1	AR	MDC1A 607855	6q22.33	<i>LAMA2</i> 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	<i>COL6A1</i> 120220	Collagen type VI subunit alpha 1	Jöbsis et al. (1996)	allelic to LGMDD5 and LGMDR22 (group 1) and UCMD1 (group 2)
Bethlem myopathy 1	2.3	AD	BTHLM1 158810	21q22.3	<i>COL6A2</i> 120240	Collagen type VI subunit alpha 2	Jöbsis et al. (1996)	allelic to LGMDD5 and LGMDR22 (group 1) and UCMD1 and Congenital myosclerosis (group 2)
Bethlem myopathy 1	2.4	AD	BTHLM1 158810	2q37.3	<i>COL6A3</i> 120250	Collagen type VI subunit alpha 3	Speer et al. (1996) Bertini et al. (1998) Pan et al. (1998)	allelic to LGMDD5 and LGMDR22 (group 1); UCMD1 (group 2)
Bethlem myopathy 1	2.5	AR	BTHLM1 158810	21q22.3	<i>COL6A2</i> 120240	Collagen type VI subunit alpha 2	Gualandi et al. (2009)	allelic to LGMDD5 and LGMDR22 (group 1) and UCMD1 and Congenital myosclerosis (group 2)
Ullrich congenital muscular dystrophy 1	2.6	AR	UCMD1 254090	21q22.3	<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)
Rigid spine syndrome 1	2.13	AR	RSMD1 602771	1p36.11	<i>SELENON</i> (formerly <i>SEPN1</i> ) 606210	Selenoprotein N	Moghadasszadeh et al. (1998, 2001) Ferreiro et al. (2002, 2004)	allelic to CFTD (group 3), multimincore disease (group 3), and desmin-related myopathy with Mallory bodies (group 5)
Congenital muscular dystrophy due to ITGA7 deficiency	2.14	AR	613204	12q13.2	<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. (2008)	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 EDM2, EDM3 and formerly LGMD1B (group 1), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
<b>CMD-Dyostroglycanopathies</b>								
Fukuyama congenital muscular dystrophy (FCMD)	2.18	AR	MDDGA4 253800	9q31.2	<i>FKTN</i> 607440	Fukutin	Toda et al. (1993) Kobayashi et al. (1998)	allelic to LGMDR13 (group 1), WWS/MDDGB4 (group 2), CMD1X (group 10A)
Walker-Warburg syndrome (WWS)	2.19	AR	MDDGB4 613152	9q31.2	<i>FKTN</i> 607440	Fukutin	Beltran-Valero de Bernabe (2003) Mercuri et al. (2009)	allelic to LGMDR13 (group 1) and FCMD/MDDGA4 (group 2) and CMD1X (group 10A)

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Walker-Warburg syndrome (WWS)	2.20	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 LGMDR11 (group 1), and MDDGB1 (group 2)
Walker-Warburg syndrome (WWS)	2.21	AR	MDDGA2 613150 MDDGB2 613156	14q24.3	<i>POMT2</i> 607439	Protein O-mannosyl transferase 2	van Reeuwijk et al. (2005) Mercuri et al. (2009)	allelic to LGMDR14 (group 1) and MDDGA2/MDDGB2/MEB (group 2)
Walker-Warburg syndrome (WWS)	2.22	AR	MDDGA5 613153	19q13.32	<i>FKRP</i> 606596	Fukutin related protein	Beltran-Valero De Bernabe et al. (2004)	allelic to LGMDR9 (group 1), MDDGB5 (group 2), MDDGA5/MEB (group 2)
Walker-Warburg syndrome (WWS)	2.23	AR	MDDGA3 253280 MDDGB3 613151	1p34.1	<i>POMGNT1</i> 606822	Protein O-mannose beta1,2-N-acetylglucosaminyl transferase	Taniguchi et al. (2003) Mercuri et al. (2009)	allelic to LGMDR15 (group 1) and MDDGA3/MEB (group 2)
Walker-Warburg syndrome (WWS)	2.24	AR	MDDGA7 614643	7p21.2	<i>ISPD</i> 614631	Isoprenoid synthase domain containing	Roscioli et al. (2012) Willer et al. (2012)	Allelic to LGMDR20 (group 1)
Walker-Warburg syndrome (WWS)	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 LGMDR24 (group 1)
Walker-Warburg syndrome (WWS)	2.26	AR	MDDGA13 615287	11q13.2	<i>B4GAT1</i> 605517	Beta-1,4-glucuronyl transferase 1 (Beta-1,3-N-acetylglucosaminyltransferase 1)	Buyse et al. (2013) Shaheen et al. (2013)	
Muscle-eye-brain disease (MEB)	2.27	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 LGMDR15 (group 1) and MDDGA3/WWS (group 2)
Muscle-eye-brain disease (MEB)	2.28	AR	MDDGA5 613153	19q13.32	<i>FKRP</i> 606596	Fukutin related protein	Beltran-Valero De Bernabe et al. (2004)	allelic to LGMDR9 (group 1), MDDGB5 (group 2), MDDGA5/WWS (group 2)
Muscle-eye-brain disease (MEB)	2.29	AR	MDDGA2 613150 MDDGB2 613156	14q24.3	<i>POMT2</i> 607439	Protein O-mannosyl transferase 2	Mercuri et al. (2006)	allelic to LGMDR14 (group 1) and MDDGA2/MDDGB2/WWS (group 2)
Muscle-eye-brain disease (MEB)	2.30	AR	MDDGA14 615350	3p21.31	<i>GMPPB</i> 615320	GDP-mannose pyrophosphorylase B	de Carss et al. (2013)	allelic to LGMDR19 (group 1), MDDGB14 (group 2), congenital myasthenic syndrome (group 11)
Muscle-eye-brain disease (MEB)	2.31	AD	MEB	11q13.2q14.1	<i>11q13.2q14.1 duplication</i>		Villar-Quiles et al. (2020)	
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B1	2.32	AR	MDDGB1 607423	9q34.13	<i>POMT1</i> 607423	Protein-O-mannosyl transferase 1	van Reeuwijk et al. (2006) Mercuri et al. (2009)	allelic to LGMDR11 (group 1) and MDDGA1 (group 2)
Muscular dystrophy-dystroglycanopathy, congenital with or without mental retardation (formerly MDC1C)	2.33	AR	MDDGB5 606612	19q13.32	<i>FKRP</i> 606596	Fukutin related protein	Brockington et al. (2001b) Topaloglu et al. (2003) Mercuri et al. (2009)	allelic to LGMDR9 (group 1), MDDGA5/WWS (group 2), MDDGA5/MEB (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.34	AR	MDDGA6 613154 MDDGB6 608840	22q12.3	<i>LARGE1</i> 603590	Acetylglucosaminyl-transferase-like protein (like-glycosyl transferase)	Longman et al. (2003) Mercuri et al. (2009)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.35	AR	CDG1E 608799	20q13.13	<i>DPM1</i> 603503	Dolichyl-phosphate mannosyltransferase 1, catalytic subunit	Yang et al. (2013)	
Congenital disorder of glycosylation type 1	2.36	AR	CDG1F 609180	17p13.1	<i>MPDU1</i> 604041	Mannose-P-dolichol utilization defect 1	van Tol et al. (2019)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and severe epilepsy	2.37	AR	CDG1U 615042	9q34.11	<i>DPM2</i> 603564	Dolichyl-phosphate mannosyltransferase polypeptide 2, regulatory subunit	Barone et al. (2012)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.38	AR	MDDGA9 616538	3p21.31	<i>DAG1</i> 128239	Dystrophin-Associated Glycoprotein 1	Geis et al. (2013)	allelic to LGMDR16 (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A10	2.39	AR	MDGGA10 615041	12q14.2	<i>RXYLT1</i> 605862	Ribitol xylosyltransferase 1 (transmembrane protein 5)	Vuillaumier-Barrot et al. (2013)	

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Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.40	AR	MDDGA11 615181	1q42.3	<i>B3GALNT2</i> 610194	Beta-1,3-N-acetylgalactosaminyl-transferase 2	Stevens et al. (2013)	
WWWS/MEB like Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.41	AR	MDDGA12 615249	8p11.21	<i>POMK</i> 615247	Protein-O-mannose kinase	Jae et al. (2013)	Allelic to MDDGC12 (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and mental retardation	2.42	AR	MDDGB14 615351	3p21.31	<i>GMPPB</i> 615320	GDP-mannose pyrophosphorylase B	Carss et al. (2013)	allelic to LGMDR19 (group 1), MEB/MDDGA14 (group 2) and congenital myasthenic syndrome (group 11)
Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by <i>TRAPPC11</i> mutations	2.43	AR		4q35.1	<i>TRAPPC11</i> 614138	Trafficking protein particle complex 11	Liang et al. (2015) Larson et al. (2018)	allelic to LGMDR18 (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and epilepsy	2.44	AR		17q21.32	<i>GOSR2</i> 604027	Golgi SNAP receptor complex member 2	Larson et al. (2018)	
<b>Other congenital muscular dystrophies</b>								
Congenital muscle dystrophy with joint hyperlaxity	2.45	AR		3p23-21	?		Tetreault et al. (2006)	
Congenital muscle dystrophy with mitochondrial structural abnormalities (megaconial type)	2.46	AR	MDCMC 602541	22q13.33	<i>CHKB</i> 612395	Choline kinase beta	Mitsuhashi et al. (2011)	
Congenital muscular dystrophy	2.47	AR	MDC1B 604801	1q42	?		Brockington et al. (2000)	
Congenital muscular dystrophy with rigid spine related to <i>ACTA1</i>	2.48	AR	Possibly identical to MDC1B 604801	1q42.13	<i>ACTA1</i> 102610	Alpha actin, skeletal muscle	O'Grady et al. (2014)	allelic to NEM3, CFTD, Cap myopathy (group 3)
<i>GOLGA2</i> -related congenital muscle dystrophy with brain involvement	2.49	AR		9q34.11	<i>GOLGA2</i> 602580	Golgin A2	Shamseldin et al. (2016)	
Muscular dystrophy, congenital Davignon-Chauveau type	2.50	AR	MDCDC 617066	15q22.31	<i>TRIP4</i> 604501	Thyroid hormone receptor interactor 4	Davignon et al. (2016)	allelic to SMABF1 (group 12)
Muscular dystrophy, congenital, with cataracts and intellectual disability	2.51	AR	MDCCAID 617404	17p13.3	<i>INPP5K</i> 607875	Inositol Polyphosphate-5-Phosphatase K	Osborn et al. (2017) Wiessner et al. (2017)	
Congenital muscular dystrophy related to <i>MSTO1</i>	2.52	AR		1q22	<i>MSTO1</i> 617619	Misato 1, mitochondrial distribution and morphology regulator (M)	Nasca et al. (2017) Ardicli et al. (2019) Donkervoort et al. (2019)	allelic to MMYAT (group 13 and 16)
Congenital muscular dystrophy related to <i>RYR1</i>	2.53	AR		19q13.2	<i>RYR1</i> 180901	Ryanodine receptor	Helbling et al. (2019)	allelic to CFTD, CNM related to RYR1, CCD, CNMDU1, minicore myopathy with external ophthalmoplegia, DuCD (group 3), Late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to <i>RYR1</i> (group 16)

**GROUP 3. CONGENITAL MYOPATHIES**

<b>Nemaline myopathies</b>								
Nemalin myopathy 1	3.1	AD	NEM1 609284	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 CFTD, CAPM1 (group 3)

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Nemalin myopathy 2	3.2	AR	NEM2 256030	2q23.3	<i>NEB</i> 161650	Nebulin	Wallgren-Pettersson et al. (1995, 2002) Pelin et al. (1999) Lehtokari et al. (2006) Nowak et al. (1999)	allelic to Distal myopathy with nebulin defect (group 4)
Nemalin myopathy 3	3.3	AD	NEM3 161800	1q42.13	<i>ACTA1</i> 102610	Actin, alpha 1, skeletal muscle	Nowak et al. (1999)	allelic to CMD with rigid spine (group 2), CFTD and cap myopathy (group 3)
Nemalin myopathy 4	3.4	AD	NEM4 609285	9p13.3	<i>TPM2</i> 190990	Tropomyosin 2 (beta)	Donner et al. (2002) Monnier et al. (2009)	allelic to CAPM2 (group 3), DA2B and DA1A (group 16)
Nemalin myopathy 5	3.5	AR	NEM5 605355	19q13.42	<i>TNNT1</i> 191041	Troponin T type 1 (skeletal, slow)	Johnston et al. (2000)	
Nemalin myopathy 6	3.6	AD	NEM6 609273	15q22.31	<i>KBTD13</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>KBTD13</i> (group 1)
Nemalin myopathy 7	3.7	AR	NEM7 610687	14q13.1	<i>CFL2</i> 601443	Cofilin 2 (muscle)	Agrawal et al. (2007)	
Nemalin myopathy 8	3.8	AR	NEM8 615348	3p22.1	<i>KLHL40</i> 615340	Kelch-like family member 40	Ravenscroft et al. (2013)	
Nemalin myopathy 9	3.9	AR	NEM9 615731	2q31.1	<i>KLHL41</i> 607701	Kelch-like family member 41	Gupta et al. (2013)	
Nemalin myopathy 10	3.10	AR	NEM 10 616165	3p14.1	<i>LMOD3</i> 616112	Leiomodin 3	Yuen et al. (2014)	
Nemalin myopathy 11	3.11	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 Nemalin myopathy and facial dysmorphism	3.12	AR	KFS4 616549	22q12.1	<i>MYO18B</i> 607295	Myosin XVIIIIB	Malfatti et al. (2015)	
Myopathy with nemaline bodies	3.13	AR		15q13-q14	<i>RYR3</i> 180903	Ryanodine receptor 3	Nilipour et al. (2018)	
<b>Other congenital myopathies</b>								
Myopathy, congenital, with fiber-type disproportion 1	3.14	AD, AR	CFTD 255310	1q42.13	<i>ACTA1</i> 102610	Actin, alpha 1, skeletal muscle	Clarke et al. (2003) Laing et al. (2004)	allelic to CMD with rigid spine (group 2), NEM3 and cap myopathy (group 3)
Myopathy, congenital, with fiber-type disproportion	3.15	AD, AR	CFTD 255310	1p36.11	<i>SELENON</i> (formerly <i>SEPN1</i> ) 606210	Selenoprotein N	Clarke et al. (2006)	allelic to RSMD1 (group 2), CFTD (group 3), Multiminicore disease (group 3), <i>DES</i> -related myopathy with Mallory bodies (group 5)
Myopathy, congenital, with fiber-type disproportion	3.16	AD, AR	CFTD 255310	1q21.3	<i>TPM3</i> 191030	Tropomyosin 3	Clarke et al. (2008)	allelic to NEM1, CAPM1 (group 3)
Myopathy, congenital, with fiber-type disproportion	3.17	AR	CFTD 255310	19q13.2	<i>RYR1</i> 180901	Ryanodine receptor	Clarke et al. (2010)	allelic to CMD related to <i>RYR1</i> (group 2), CNM related to <i>RYR1</i> , CCD, CNMDU1, minicore myopathy with external Ophthalmoplegia (group 3), Late onset axial myopathy (group 5), MHS1 (group 8) Fetal akinesia related to <i>RYR1</i> (group 16)
Myopathy congenital, with fiber-type disproportion	3.18	AD	CFTD 255310	14q11.2	<i>MYH7</i> 160760	Myosin, heavy chain 7, cardiac muscle, beta	Ortolano et al. (2011)	allelic to MSMA and MSMB (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10A)
Myopathy congenital, with fiber-type disproportion	3.19	AR	CFTD 255310	12q24.11	<i>MYL2</i> 160781	Myosin regulatory light chain 2	Waterman et al. (2013)	Allelic to CMH10 (group 10A)
Myopathy congenital, with fiber-type disproportion	3.20	AR	CNM6 617760	2q31.1	<i>ZAK</i> 609479	Leucine zipper-and sterile alpha motif-containing kinase	Vasli et al. (2017)	
Myotubular myopathy, X-linked	3.21	XR	CNMX 310400	Xq28	<i>MTM1</i> 300415	Myotubularin 1	Thomas et al. (1987) Laporte (1996, 1997, 2000)	
Centronuclear myopathy 1	3.22	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)
Centronuclear myopathy 2	3.23	AR, AD	CNM2 255200	2q14.3	<i>BINI</i> 601248	Bridging integrator 1 (Amphiphysin)	Nicot et al. (2007) Böhm et al. (2014)	

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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)
Centronuclear myopathy related to <i>RYR1</i>	3.24	AR		19q13.2	<i>RYR1</i> 180901	Ryanodine receptor	Wilmshurst et al. (2010)	allelic to CMD related to <i>RYR1</i> (group 2), CFTD, CCD, minicore myopathy with external Ophthalmoplegia, CNMDU1, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to <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 LGMDR10 (group 1), MmD related to <i>TTN</i> (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A), LCCS related to <i>TTN</i> (group 12)
Centronuclear myopathy 5	3.26	AR	CNM5 615959	2q35	<i>SPEG</i> 615950	SPEG complex locus	Agrawal et al. (2014)	
Centronuclear myopathy 4	3.27	AD	CNM4 614807	16p13.3	<i>CCDC78</i> 614666	Coiled-coil domain-containing protein 78	Majcenko et al. (2012)	
Central core disease, dominant	3.28	AD	CCD 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 CMD related to <i>RYR1</i> (group 2), CFTD, CNM related to <i>RYR1</i> , minicore myopathy with external ophthalmoplegia, CNMDU1, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to <i>RYR1</i> (group 16)
Central core disease, recessive (transient multiminicore myopathy)	3.29	AR	CCD 117000	19q13.2	<i>RYR1</i> 180901	Ryanodine receptor	Ferreiro et al. (2002a) Jungbluth et al. (2002)	allelic to CMD related to <i>RYR1</i> (group 2), CFTD, CNM related to <i>RYR1</i> , minicore myopathy with external ophthalmoplegia, CNMDU1 (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to <i>RYR1</i> (group 16)
Minicore myopathy with external ophthalmoplegia	3.30	AR	255320	19q13.2	<i>RYR1</i> 180901	Ryanodine receptor	Monnier et al. (2003) Jungbluth et al. (2005)	allelic to CMD related to <i>RYR1</i> (group 2), CFTD, CNM related to <i>RYR1</i> , CCD, CNMDU1, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to <i>RYR1</i> (group 16)
Multiminicore disease (MmD), classical form	3.31	AR	255320	1p36.11	<i>SELENON</i> (formerly <i>SEPN1</i> ) 606210	Selenoprotein N	Ferreiro et al. (2002) Ferreiro et al. (2004)	allelic to RSMD1 (group 2), CFTD (group 3), <i>DES</i> -related myopathy with Mallory bodies (group 5)
Multiminicore disease (MmD) related to <i>TTN</i>	3.32	AR		2q31.2	<i>TTN</i> 188840	Titin	Ge et al. (2019)	allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A), LCCS related to <i>TTN</i> (group 12)
Early onset myopathy, areflexia, respiratory distress and dysphagia	3.33	AR	EMARDD 614399	5q23.2	<i>MEGF10</i> 612453	Multiple EGF-like-domains 10	Logan et al. (2011)	Allelic to recessive congenital myopathy with minicores (group 3)
Recessive congenital myopathy with minicores	3.34	AR	EMARDD 614399	5q23.2	<i>MEGF10</i> 612453	Multiple EGF-like-domains 10	Boyden et al. (2012)	Allelic to EMARDD (group 3)
Myopathy, myosin storage, autosomal dominant	3.35	AD	MSMA 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 CFTD, MSMB, SPMM (group 3); MPD1 (group 4); CMH1, CMD1S and LVNC5 (group 10A)
Myopathy, myosin storage, autosomal recessive	3.36	AR	MSMB 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 CFTD, MSMA and SPMM (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10A)
Myopathy, proximal, and ophthalmoplegia (inclusion body myopathy 3)	3.37	AD, AR	MYPOP 605637	17p13.1	<i>MYH2</i> 160740	Myosin, heavy chain 2, skeletal muscle, adult	Martinsson et al. (1999, 2000) Tajsharghi et al. (2010)	
Isolated inclusion body myopathy	3.38	AD	IBMPFD3 615424	12q13.13	<i>HNRNP1A1</i> 164017	Heterogeneous nuclear ribonucleoprotein A1	Izumi et al. (2015)	allelic to ALS20 (group 12)

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Cap myopathy CAPM1	3.39	AD, AR	NEM1 609284	1q21.3	<i>TPM3</i> 191030	Tropomyosin 3	De Paula et al. (2009) Ohlsson et al. (2009)	allelic to NEM1, CFTD (group 3)
Cap myopathy CAPM2	3.40	AD	NEM4 609285	9p13.3	<i>TPM2</i> 190990	Tropomyosin 2, beta	Tajsharghi et al. (2007) Lehtokari et al. (2007)	allelic to NEM4 (group 3), DA1A and DA2B (group 16)
Cap myopathy	3.41	AD		1q42.13	<i>ACTA1</i> 102610	Actin, alpha 1, skeletal muscle	Hung et al. (2010)	allelic to CMD with rigid spine (group 2), CFTD and NEM3 (group 3)
Congenital neuromuscular disease with uniform type I fiber (CNMDU1)	3.42	AR, AD	CCD 117000	19q13.2	<i>RYR1</i> 180901	Ryanodine receptor I	Sato et al. (2007)	allelic to CMD related to <i>RYR1</i> (group 2), CFTD, CNM related to <i>RYR1</i> , CCD, minicore myopathy with external ophthalmoplegia, DuCD (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to <i>RYR1</i> (group 16)
Salih myopathy, (Congenital myopathy with fatal cardiomyopathy)	3.43	AR	SALMY 611705	2q31.2	<i>TTN</i> 188840	Titin	Carmignac et al. (2007)	Allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> (group 3), MmD related to <i>TTN</i> (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A)
Congenital skeletal myopathy and fatal cardiomyopathy	3.44	AR		11p11.2	<i>MYBPC3</i> 600958	Cardiac myosin binding protein-C	Tajsharghi et al. (2010)	allelic to CMH4, CMD1MM and LVNC10 (group 10A)
Congenital myopathy Compton-North	3.45	AR	MYPCN 612540	12q12	<i>CNTN1</i> 600016	Contactin-1	Compton et al. (2008)	
Sarcotubular myopathy	3.46	AR		9q33.1	<i>TRIM32</i> 602290	Tripartite motif containing 32	Schooser et al. (2005)	allelic to LGMDR8 (group 1)
Congenital myopathy related to <i>HACD1</i>	3.47	AR		10p12.33	<i>HACD1</i> ( <i>PTPLA</i> ) 610467	3-Hydroxyacyl-CoA dehydratase (Protein tyrosine phosphatase-like)	Muhammad et al. (2013)	allelic to ARVD6 (group 10B)
Congenital myopathy with ophthalmoplegia related to <i>CACNA1S</i>	3.48	AR		1q32.1	<i>CACNA1S</i> 114208	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Hunter et al. (2015)	Allelic to HOKPP1 (group 7) MHS5 (group 8)
Congenital myopathy with neuropathy and deafness	3.49	AR	NEDHND 617519	19q13.2	<i>SPTBN4</i> 606214	Spectrin, beta, nonerythrocytic, 4	Knierim et al. (2017)	
Myopathy, congenital, with excess of muscle spindles	3.50	AD	CMEMS 218040	11p15.5	<i>HRAS</i> 190020	V-Ha-RAS Harvey rat sarcoma viral oncogene homolog	Quélin et al. (2017)	
Carey-Fineman-Ziter syndrome (formerly congenital myopathy with Moebius sequence and Robin sequence)	3.51	AR	CFZS 254940	9q34.2	<i>MYMK</i> 615345	Myomaker	Di Gioia et al. (2017)	
Myopathy, congenital, Baily-Bloch (Native American myopathy)	3.52	AR	MYPBB 255995	12q13.3	<i>STAC3</i> 615521	SH3 and cysteine-rich domains 3	Horstick et al. (2013)	Allelic to Myopathy, congenital, with malignant hyperthermia susceptibility (group 3)
Myopathy, congenital, with malignant hyperthermia susceptibility	3.53	AR		12q13.3	<i>STAC3</i> 615521	SH3 and cysteine-rich domains 3	Zaharieva et al. (2018)	Allelic to MYPBB (group 3)
Myopathy congenital with structured cores and Z-line abnormalities	3.54	AD	MYOCOZ 618654	1q43	<i>ACTN2</i> 102573	Actinin, alpha 2	Lornage et al. (2019)	Allelic to MPD6 (group 4), CMH23 and CMD1AA (group 10A)
Myopathy congenital with fast twitch (type II) fiber atrophy	3.55	AR	MYOFTA 618414	2q34	<i>MYL1</i> 160780	Myosin, light polypeptide 1, alkali, skeletal fast	Ravenscroft et al. (2018)	
Congenital amyotrophy	3.56	AR		16p13.3	<i>CACNA1H</i> 607904	Calcium channel, voltage-dependent, T type, alpha-1H subunit	Carter et al. (2019)	
Congenital multi-minicore myopathy	3.57	AR		3q26.33	<i>FXR1</i> 600819	FMR1 autosomal homolog	Estan et al. (2019)	
Congenital Myopathy related to PAX7	3.58	AR		1p36.13	<i>PAX7</i> 167410	Paired Box gene 7	Feichtinger et al. (2019)	
Congenital Myopathy related to <i>SCN4A</i>	3.59	AR		17q23.3	<i>SCN4A</i> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Zaharieva et al. (2016) Gonorazky et al. (2017) Sloth et al. (2018)	allelic to HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16)

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Congenital Myopathy related to <i>PYROXD1</i>	3.60	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)
Congenital Myopathy related to <i>TNPO3</i>	3.61	AD		7q32.1	<i>TNPO3</i> 610032	Transportin 3	Vihola et al. (2019) Angelini et al. (2019)	allelic to LGMDD2 (group 1)
Scapuloperoneal myopathy, <i>MYH7</i> -related	3.62	AD	SPMM 181430	14q11.2	<i>MYH7</i> 160760	Myosin, heavy chain 7, cardiac muscle, beta	Pegoraro et al. (2007)	Allelic to CFTD, MSMA and MSMB (group 3); MPD1 (group 4); CMH1, CMD1S and LVNC5 (group 10A)
Congenital myopathy related to <i>UNC45B</i>	3.63	AR		17q12	<i>UNC45B</i> 611220	UNC45 Myosin Chaperone B	Dafsari et al. (2019)	
Dusty core disease related to RYR1	3.64	AR	DuCD	19q13.2	<i>RYR1</i> 180901	Ryanodine receptor I	Garibaldi et al. (2019)	allelic to CMD related to RYR1 (group 2), CFTD, CNM related to RYR1, CCD, minicore myopathy with external ophthalmoplegia, CNMDU1 (group 3), late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia related to RYR1 (group 16)

**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)
Tibial muscular dystrophy (Udd)	4.2	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 SALMY (group 3), MFM9 (group 5), CMH9 (group 10A), CMD1G (group 10A)
Nonaka Myopathy	4.3	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)	
Distal myopathy 1 (Laing)	4.4	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, MSMA, MSMB and SPMM (group 3), CMH1, CMD1S and LVNC5 (group 10A)
Vocal cord and pharyngeal distal myopathy (VCPDM) reclassified as ALS21	4.5	AD	ALS21 606070	5q31.2	<i>MATR3</i> 164015	Matrin 3	Feit et al. (1998) Senderek et al. (2009)	Allelic to ALS21 (group 12)
Myopathy distal 3	4.6	AD	MPD3 610099	8p22-q11 12q13-q22	?	?	Haravuori et al. (2004)	
Welander distal myopathy	4.7	AD	WDM 604454	2p13.3	<i>TIA1</i> 603518	Cytotoxic granule-associated RNA binding protein	Ahlberg et al. (1999) Hackman et al. (2013) Klar et al. (2013)	allelic to Welander-like syndrome (group 4)
Welander-like distal myopathy	4.8	Digenic		5q35.3 2p13.3	<i>SQSTM1</i> 601530 + <i>TIA1</i> 603518	Sequestosome 1 + Cytotoxic granule-associated RNA binding protein	Lee et al. (2018)	<i>SQSTM1</i> Allelic to DMRV (group 4) and FTDALS3 (group 12)  <i>TIA1</i> Allelic to WDM (group 4)
Vacuolar neuromyopathy	4.9	AD	601846	19p13.3	?	?	Servidei et al. (1999)	
Distal myopathy with myotilin defect	4.10	AD		5q31.2	<i>MYOT</i> 604103	Myotilin	Penisson-Besnier et al. (1998, 2006)	allelic to formerly LGMD1A (group 1), MFM3 (group 5), spheroid body myopathy (group 5), LVHT/NC (group 10A)
Distal myopathy with nebulin defect	4.11	AR		2q23.3	<i>NEB</i> 161650	Nebulin	Wallgren-Pettersson et al. (2007)	allelic to NEM2 (group 3) and AD Distal myopathy with nebulin defect (group 4)
Distal myopathy with nebulin defect	4.12	AD		2q23.3	<i>NEB</i> 161650	Nebulin	Kiiski et al. (2019)	allelic to NEM2 (group 3) and AR Distal myopathy with nebulin defect (group 4)
Distal myopathy, Tateyama type	4.13	AD	MPDT 614321	3p25.3	<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)

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Late onset distal myopathy (Markesbery-Griggs)	4.14	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)
Miyoshi muscular dystrophy 3	4.15	AR	MMD3 613319	11p14.3	<i>AN05</i> 608662	Anoctamin 5	Bolduc et al. (2010)	allelic to LGMDR12 (group 1)
Distal myopathy related to <i>DNM2</i>	4.16	AD		19p13.2	<i>DNM2</i> 602378	Dynamin 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	4.17	AD		9p21.3	<i>KLHL9</i> 611201	Kelch-like 9	Cirak et al. (2010)	
Distal Myopathy 4	4.18	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)
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 1	4.19	AD	IBMPFD1 167320	9p13.3	<i>VCP</i> 601023	Valosin-containing protein	Palmio et al. (2011)	allelic to Scapuloperoneal myopathy (group 1), IBMPFD1 (group 5), ALS14 (group 12) and CMT2Y (group 14)
Distal myopathy 5	4.20	AR	MPD5 617030	14q32.33	<i>ADSSL1</i> 612498	Adenylosuccinate synthase-like	Park et al. (2016)	
Myopathy, distal, with rimmed vacuoles	4.21	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	4.22	AD		7q36.3	<i>DNAJB6</i> 611332	Hsp40 homologue, subfamily B, number 6	Ruggieri et al. (2015)	allelic to LGMD1 (group 1) and distal myopathy (group 4)
Distal myopathy	4.23	AD		7q36.3	<i>DNAJB6</i> 611332	Hsp40 homologue, subfamily B, number 6	Palmio et al. (2019)	Allelic to LGMD1 (group 1), Rimmed vacuole distal myopathy (group 4)
Rimmed vacuole myopathy	4.24	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) HMN2A (group 12) and CMT2L (group 14)
Distal Myopathy 6, Adult-onset	4.25	AD	MPD6 618655	1q43	<i>ACTN2</i> 102573	Actinin alpha-2	Savarese et al. (2019)	Allelic to MYOCOZ (group 3), CMH23 and CMD1AA (group 10A)
Distal Myopathy and motor neuropathy	4.26	AD		12q24.23	<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

*Myofibrillar myopathies*

Myofibrillar myopathy 1	5.1	AD	MFM1 601419	2q35	<i>DES</i> 125660	Desmin	Goldfarb et al. (1998) Munoz-Marmol et al. (1998)	allelic to formerly LGMD1 related to <i>DES</i> and formerly LGM2R (group1), MFM1 with ARCV7 (group 5), CMD11 (group 10A) and ARVC7 (group 10B)
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 CMDIII (group 10A)
Myofibrillar myopathy 3	5.3	AD	MFM3 609200	5q31.2	<i>MYOT</i> 604103	Myotilin	Selcen and Engel (2004)	allelic to formerly LGMD1A (group 1), distal myotilinopathy (group 4), spheroid body myopathy (group 5), LVHT/NC (group 10A)
Spheroid body myopathy	5.4	AD	182920	5q31.2	<i>MYOT</i> 604103	Myotilin	Foroud et al. (2005)	allelic to formerly LGMD1A (group 1), distal myotilinopathy (group 4), MFM3 (group 5)
Myofibrillar myopathy 4	5.5	AD	MFM4 609452	10q23.2	<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	MFM5 609524	7q32.1	<i>FLNC</i> 102565	Filamin C (gamma)	Vorgerd et al. (2005)	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 CMD1HH (group 10A) and CMT related to BAG3 (group 14)
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)	

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Myofibrillar myopathy 8	5.9	AR	MF8 617258	12p12.1	<b>PYROXD1</b> 617220	Pyridine nucleotide-disulphide oxidoreductase domain 1	O'Grady et al. (2016)	allelic to LGMD related to <b>PYROXD1</b> (group 1) and Congenital Myopathy related to <b>PYROXD1</b> (group 3)
Myofibrillar myopathy 9 with early respiratory failure	5.10	AD	MF9 (HMERF) 603689	2q31.2	<b>TTN</b> 188840	Titin	Nicolao et al. (1999) Lange et al. (2005)	allelic to LGMDR10 (group 1), CMN related to <b>TTN</b> (group3), MmD related to <b>TTN</b> (group 3), SALMY (group 3), TMD (group 4), CMH9 (group 10A), CMD1G (group 10A), LCCS related to <b>TTN</b> (group 12)
Myofibrillar myopathy 10	5.11	AR	MF10 619040	10p11.23	<b>SVIL</b> 604126	Supervillin	Hedberg-Oldfors et al. (2020)	
Desmin-related myopathy with Mallory bodies	5.12	AD	RSMD1 602771	1p36.11	<b>SELENON</b> (formerly <b>SEPN1</b> ) 606210	Selenoprotein N	Ferreiro et al. (2004)	allelic to RSMD1 (group 2), CFTD (group 3) Multiminicore disease (group 3)
Cardiac and skeletal aggregate myopathy	5.13	Digenic		1p36.11 2p.23.3	<b>TRIM63</b> 06131 + <b>TRIM54</b> 606474	Tripartite motif containing 63 (MURF1)+Ring finger protein 30 (MURF3)	Olivé et al. (2015)	
Myofibrillar myopathy with arrhythmogenic right ventricular cardiomyopathy (ARCV7) also listed in group 10B	5.14	AD	MF1 601419	10q22 > 2q35	<b>DES</b> 125660	Desmin	Melberg et al. (1999) Kuhl et al. (2008) Hedberg et al. (2012)	allelic to formerly LGMD1 related to <b>DES</b> and formerly LGMD2R (group 1), MF1 (group 5), CMD11 (group 10A) and ARVC7 (group 10B)
<b>Miscellaneous</b>								
Danon disease	5.15	XD	GSD2B 300257	Xq24	<b>LAMP2</b> 309060	Lysosomal-associated membrane protein 2	Nishino et al. (2000) Musumeci et al. (2005)	
Myopathy with excessive autophagy	5.16	XR	MEAX 310440	Xq28	<b>VMA21</b> 300913	S. Cerevisiae homolog of VMA21	Saviranta et al. (1988) Villard et al. (2000) Minassian et al. (2002) Munteanu et al. (2008) Ramachandran et al. (2013) Crockett et al. (2014) Cortese et al. (2014)	
Autophagic vacuolar myopathy	5.17	AR	CLN3 204200	16p12.1	<b>CLN3</b> 607042	Ceroid-lipofuscinosis, neuronal 3 (= Battenin)		
Oculopharyngeal muscular dystrophy	5.18	AD	OPMD 164300	14q11.2	<b>PABPN1</b> 602279	Polyadenylate-binding protein, nuclear 1	Brais et al. (1995, 1998) Robinson et al. (2005)	
Oculopharyngodistal myopathy 1	5.19	AD	OPDM1 164310	8q22.3	<b>LRP12</b> 618299	Low density lipoprotein receptor-related protein 12	Ishiura et al. (2019)	
Oculopharyngodistal myopathy 2	5.20	AD	OPDM2 618940	19p13.12	<b>GIPCI</b> 605072	GIPC PDZ Domain-containing family, member 1	Deng et al. (2020)	
Epidermolysis bullosa simplex associated with muscular dystrophy	5.21	AR	EBSMD 226670	8q24.3	<b>PLEC</b> 601282	Plectin	Gache et al. (1996) Smith et al. (1996) Wuyts et al. (1996)	allelic to LGMDR17 (group 1), LGMD with Ophthalmoplegia (group 1), myasthenic syndrome related to <b>PLEC</b> (group 11)
Muscle hypertrophy	5.22	AR	MSLHP 614160	2q32.2	<b>MSTN</b> 601788	Myostatin	Schuelke et al. (2004)	
Fibrodysplasia ossificans progressiva	5.23	AD	FOP 135100	2q24.1	<b>ACVRI</b> 102576	Activin A receptor, type 1	Shore et al. (2006)	
Creatine phosphokinase, elevated serum (formerly HyperCKemia, idiopathic)	5.24	AD	123320	3p25.3	<b>CAV3</b> 601253	Caveolin-3	Carbone et al. (2000)	allelic to formerly LGMD1C (group 1), MPDT (group 4), RMD2 (group 6), CMH1 and LQT9 (group 10)
X-linked myopathy with postural muscle atrophy	5.25	XR	XMPMA 300696	Xq26.3	<b>FHL1</b> 300163	Four-and-a-half LIM domains 1	Windpassinger et al., 2008	allelic to EDMD6 (group 1), RBMX1A/B (group 5), SPM (group 5)
Scapuloperoneal myopathy	5.26	XD	SPM 300695	Xq26.3	<b>FHL1</b> 300163	Four-and-a-half LIM domains 1	Quinzii et al. (2008)	allelic to EDMD6 (group 1), RBMX1A/B (group 5), XMPMA (group 5)

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Reducing body myopathy (1A and 1B)	5.27	XD	RBMX1A 300717 RBMX1B 300718	Xq26.3	<i>FHL1</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	5.28	XR	EMWX 300211	Xp22.3	?	?	Ryan et al. (1999)	
Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia 1	5.29	AD	IBMPFD1 167320	9p13.3	<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	5.30		IBMPFD2 615422	7p15.2	<i>HNRNPA2B1</i> 600124	Heterogeneous nuclear ribonucleoprotein A2/B1	Kim et al. (2013)	
Myopathy with lactic acidosis, hereditary	5.31	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>	5.32	AD		19q13.2	<i>RYR1</i> 180901	Ryanodine receptor 1 (skeletal)	Løseth et al. (2013)	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), MHS1 (group 8), Fetal akinesia related to <i>RYR1</i> (group 16)
Tubular aggregate myopathy 1	5.33	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)	
Tubular aggregate myopathy 2	5.34	AD	TAM2 615883	12q24.31	<i>ORAI1</i> 610277	ORAI calcium release-activated calcium modulator 1	Nesin et al. (2014)	
Vacuolar myopathy with CASQ1 aggregates	5.35	AD	VMCQA 616231	1q23.2	<i>CASQ1</i> 114250	Calsequestrin 1	Rossi et al. (2014)	
Myopathy with characteristic sarcoplasmic inclusions	5.36	AD		22q12.3	<i>MB</i> 160000	Myoglobin	Olivé et al. (2019)	
<b>GROUP 6. MYOTONIC SYNDROMES</b>								
Myotonic dystrophy 1 (Steinert)	6.1	AD	DM1 160900	19q13.32	<i>DMPK</i> 605377	Dystrophin myotonic 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	<i>see under Ion channel muscle diseases (group 7)</i>					
Myotonia, recessive (Becker)	6.4	AR	<i>see under Ion channel muscle diseases (group 7)</i>					
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)	

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Brody disease	6.9	AR	601003	16p11.2	<i>ATP2A1</i> (formerly <i>SERCA1</i> ) 108730	ATPase, Ca <sup>++</sup> transporting, fast twitch 1	Odermatt et al. (1996)	
<b>Group 7. ION CHANNEL MUSCLE DISEASES</b>								
<b>Chloride channel</b>								
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)
<b>Sodium channel</b>								
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 Congenital Myopathy related to <i>SCN4A</i> (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 Congenital Myopathy related to <i>SCN4A</i> (group 3), HYPP, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16)
Hypokalaemic periodic paralysis	7.5	AD		1q23.2	<i>ATPIA2</i> 182340	ATPase, NA <sup>+</sup> /K <sup>+</sup> 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 Congenital Myopathy related to <i>SCN4A</i> (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 Congenital Myopathy related to <i>SCN4A</i> (group 3), HYPP, HOKPP2 and PMC (group 7), CMS16 (group 11), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16)
<b>Calcium channel</b>								
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 congenital myopathy with Ophthalmoplegia (group 3), MHS5 (group 8)
Acetazolamide responsive hereditary paroxysmal cerebellar ataxia (also listed in group 13 "Ataxias")	7.9	AD	APCA 108500	19p13.13	<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)
<b>Potassium channel</b>								
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)	

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Periodic paralysis, potassium sensitive cardiodyrhythmic (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>						
<b>GROUP 8. MALIGNANT HYPERTHERMIAS</b>								
Malignant hyperthermia	8.1	AD	MHS1 145600	19q13.2	<b><i>RYR1</i></b> <i>180901</i>	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) Levitt et al. (1992) Moslehi et al. (1998) Iles 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	?	?		
Malignant hyperthermia	8.3	AD	MHS3 154276	7q21-q22	?	?		
Malignant hyperthermia	8.4	AD	MHS4 600467	3q13.1	?	?	Sudbrak et al. (1995)	
Malignant hyperthermia	8.5	AD	MHS5 601887	1q32.1	<b><i>CACNAIS</i></b> <i>114208</i>	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Monnier et al. (1997)	allelic to congenital myopathy with ophthalmoplegia (group 3), HOKPP1 (group 7)
Malignant hyperthermia	8.6	AD	MHS6 601888	5p	?	?	Robinson et al. (1997)	
<b>GROUP 9. METABOLIC MYOPATHIES</b>								
<b>Glycogen storage diseases</b>								
Glycogen storage disease type II, Pompe disease (also listed in group 10A)	9.1	AR	GSD2 232300	17q25.3	<b><i>GAA</i></b> <i>606800</i>	Glucosidase, alpha acid	Hers et al. (1963) Martiniuk et al. (1990) Wokke et al. (1995)	Allelic to Pompe Disease (formerly LMGD2V) (group 1)
Glycogen storage disease type III	9.2	AR	GSD3 232400	1p21.2	<b><i>AGL</i></b> <i>610860</i>	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	<b><i>GBE1</i></b> <i>607839</i>	Glycogen branching enzyme (1,4- $\alpha$ -glucan branching enzyme)	Brown et al. (1966) Bao et al. (1996)	
Glycogen storage disease type V (McArdle)	9.4	AR	GSD5 232600	11q13.1	<b><i>PYGM</i></b> <i>608455</i>	Glycogen phosphorylase, muscle	Bruno et al. (2004) 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	<b><i>PFKM</i></b> <i>610681</i>	Phosphofructokinase, muscle type	Tarui et al. (1965) Nakajima et al. (1991) Howard et al. (1996)	
Glycogen storage disease type IXd (ex type VIII)	9.6	XR	GSD9D 300559	Xq13.1	<b><i>PHKA1</i></b> <i>311870</i>	Phosphorylase kinase, alpha-1 subunit	Wehner et al. (1994)	
Glycogen storage disease type XIV (Congenital disorder of glycosylation, type It)	9.7	AR	CDG1T 614921	1p31.3	<b><i>PGM1</i></b> <i>171900</i>	Phosphoglucomutase 1	Burwinkel et al. (2004) Stojkovic et al. (2009)	
Glycogen storage disease type XV	9.8	AR	GSD15 613507	3q24	<b><i>GYGI</i></b> <i>603942</i>	Glycogenin 1	Moslemi et al. (2010)	allelic to PGBM2 (group 9)
Glycogen storage disease type 0	9.9	AR	GSD0B 611556	9q13.33	<b><i>GYS1</i></b> <i>138570</i>	Glycogen synthase 1	Kolberg et al. (2007)	
Glycogen storage disease of heart, lethal congenital	9.10	AD	261740	7q36.1	<b><i>PRKAG2</i></b> <i>602743</i>	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	<b><i>RBCK1</i></b> <i>610924</i>	RanBP-type and C3HC4-type zinc finger containing 1	Nilsson et al. (2013)	

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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)
<b>Glycolytic pathway</b>								
Phosphoglycerate kinase 1 deficiency	9.13	XR	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) Scrabble 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)	
<b>Disorders of lipid metabolism</b>								
Carnitine palmitoyl-transferase II deficiency, myopathic, stress induced	9.18	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)	
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 (VLCAD deficiency)	9.24	AR	ACADVLD 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 due to ACAD9 deficiency	9.25	AR	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	NLSMD 610717	11p15.5	<i>PNPLA2</i> 609059	Patatin-like phosphorylase domain-containing protein 2 (Adipose triglyceride lipase)	Fischer et al. (2007)	
Acute Recurrent myoglobinuria	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)	

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<b>GROUP 10A. HEREDITARY CARDIOMYOPATHIES non arrhythmogenic</b>								
<i>Hypertrophic cardiomyopathies</i>								
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, MSMA, MSMB and SPMM (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> (group3), MmD related to <i>TTN</i> (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMD1G (group 10A), LCCS related to <i>TTN</i> (group 12)
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 CFTD (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 CMD1P (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, 22	10.23	AD	CMH22 615248	10q21.3	<i>MYPN</i> 608517	Myopalladin	Purevjav et al. (2012)	allelic to NEM11 (group 3), CMD1KK and RCM4 (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)
Hypertrophic cardiomyopathy, 23	10.24	AD	CMH23 612158	1q43	<i>ACTN2</i> 102573	Actinin alpha-2	Chiu C et al. (2010)	allelic to MYOCOZ (group 3), MPD6 (group 4), CMD1AA (group 10A)
Hypertrophic cardiomyopathy, 24	10.25	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	10.26	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	10.27	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	10.28	AR	CMH27 618052	15q25.3	<i>ALPK3</i> 617608	Alpha Kinase 3	Almomani et al. (2016)	
Mitochondrial complex I deficiency, nuclear type 11	10.29	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	10.30	AR	COXPD3 610505	12q14.1	<i>TSFM</i> 604723	Ts translation elongation factor, mitochondrial (M)	Smeitink et al.(2006)	
Combined oxydative phosphorylation deficiency 8	10.31	AR	COXPD8 614096	6p21.1	<i>AARS2</i> 612035	Alanyl-tRNA synthetase 2, mitochondrial (M)	Götz et al. (2011)	
Combined oxydative phosphorylation deficiency 9	10.32	AR	COXPD9 614582	3q21.1	<i>MRPL3</i> 607118	Mitochondrial ribosomal protein L3 (M)	Galmiche et al. (2011)	
Combined oxydative phosphorylation deficiency 10	10.33	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	10.34	AR	COXPD16 615395	2q36.1	<i>MRPL44</i> 611849	Mitochondrial ribosomal protein L44 (M)	Carroll et al. (2013)	
Cardio-encephalomyopathy, fatal infantile, due to cytochrome C oxidase deficiency 1	10.35	AR	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)
Cardio-encephalomyopathy, fatal infantile, due to cytochrome C oxidase deficiency 2	10.36	AR	CEMCOX2 615119	10q24.2	<i>COX15</i> 603646	Cytochrome c oxidase assembly factor Cox15 (M)	Antonicka et al. (2003)	
<b>Dilated cardiomyopathies</b>								
Dilated cardiomyopathy, 1A	10.37	AD	CMD1A 115200	1q22	<i>LMNA</i> 150330	Lamin A/C	Fatkin et al. (1999)	allelic to EDMD2, EDMD3 and formerly LGMD1B (group1), MDCL (group 2), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermatopathy #275210, MADA #248370]
Dilated cardiomyopathy, 1B	10.38	AD	CMD1B 600884	9q13	?	?	Krajinovic et al. (1995)	
Dilated cardiomyopathy, 1C, with or without LVNC	10.39	AD	CMD1C 601493	10q23.2	<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	10.40	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	10.41	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	10.42	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> (group 3), MmD related to <i>TTN</i> (group 3), SALMY (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), LCCS related to <i>TTN</i> (group 12)
Dilated cardiomyopathy, 1H	10.43	AD	CMD1H 604288	2q14-q22	?	?	Jung et al. (1999)	

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Dilated cardiomyopathy, 1I	10.44	AD	CMD1I 604765	2q35	<i>DES</i> 125660	Desmin	Li et al. (1999)	allelic to formerly LGMD1 related to <i>DES</i> and formerly LGMD2R (group 1), MFM1 with or without ARCV7 (group 5) and ARVC7 (group 10B)
Dilated cardiomyopathy, 1J	10.45	AD	CMD1J 605362	6q23.2	<i>EYA4</i> 603550	Eyes absent homolog	Schönberger et al. (2005)	
Dilated cardiomyopathy, 1K	10.46	AD	CMD1K 605582	6q12–q16	?	?	Sylvius et al. (2001)	
Dilated cardiomyopathy, 1L	10.47	AD	CMD1L 606685	5q33.2–q33.3	<i>SGCD</i> 601411	Sarcoglycan, delta	Tsubata et al. (2000)	allelic to LGMDR6 (group 1)
Dilated cardiomyopathy, 1M	10.48	AD	CMD1M 607482	11p15.1	<i>CSRP3</i> 600824	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Knoll et al. (2002)	allelic to CMH12 (group 10A)
Dilated cardiomyopathy, 1N	10.49	AD	CMD1N	17q12	<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	10.50	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	10.51	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	10.52	AD	CMD1Q 609915	7q22.3–q31.1	?	?	Schonberger et al. (2005)	
Dilated cardiomyopathy, 1R	10.53	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	10.54	AD	CMD1S 613426	14q11.2	<i>MYH7</i> 160760	Myosin heavy chain 7 (beta), cardiac muscle	Kamisago et al. (2000)	allelic to CFTD, MSMA, MSMB and SPMM (group 3), MPD1 (group 4), CMH1 and LVNC5 (group 10A)
Dilated cardiomyopathy, 1T	10.55	AD	CMD1T	12q23.1	<i>TMPO</i> 188380	Thymopoietin (lamina-associated polypeptide 2)	Taylor et al. (2005)	
Dilated cardiomyopathy, 1U	10.56	AD	CMD1U 613694	14q24.2	<i>PSEN1</i> 104311	Presenilin 1	Li et al. (2006)	
Dilated cardiomyopathy, 1V	10.57	AD	CMD1V 613697	1q42.13	<i>PSEN2</i> 600759	Presenilin 2	Li et al. (2006)	
Dilated cardiomyopathy, 1W	10.58	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	10.59	AR	CMD1X 611615	9q31.2	<i>FKTN</i> 607440	Fukutin	Murakami et al. (2006)	allelic to LGMDR13 (group 1), FCMD/MDDGA4 and WWS/MDDGB4 (group 2)
Dilated cardiomyopathy, 1Y	10.60	AD	CMD1Y 611878	15q22.2	<i>TPM1</i> 191010	Tropomyosin-1	Olson et al. (2010)	allelic to CMH3 and LVNC9 (group 10A)
Dilated cardiomyopathy, 1Z	10.61	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	10.62	AD	CMD1AA 612158	1q42–q43	<i>ACTN2</i> 102573	Actinin alpha-2	Mohapatra et al. (2003)	allelic to MYOZOZ (group 3), MPD6 (group 4), CMH23 (group 10A)
Dilated cardiomyopathy, 1BB	10.63	AD/AR	CMD1BB 612877	18q12.1	<i>DSG2</i> 125671	Desmoglein 2	Posch et al. (2008)	allelic to ARVD10 (group 10B)
Dilated cardiomyopathy, 1CC	10.64	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	10.65	AD	CMD1DD 613172	10q25.2	<i>RBM20</i> 613171	RNA-binding motif protein 20	Brauch et al. (2009)	
Dilated cardiomyopathy, 1EE	10.66	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	10.67	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	10.68	AR	CMD1GG 613642	5p15.33	<i>SDHA</i> 600857	Succinate dehydrogenase complex, subunit a, flavoprotein (M)	Levitass et al. (2010)	
Dilated cardiomyopathy, 1HH	10.69	AD	CMD1HH 613881	10q26.11	<i>BAG3</i> 603883	Bel2-associated athanogene 3	Norton et al. (2011)	allelic to MFM6 (group 5) and CMT related to BAG3 (group 14)
Dilated cardiomyopathy, 1II	10.70	AD	CMD1II 615184	11q23.1	<i>CRYAB</i> 123590	Alpha-B crystallin	Inagaki et al. (2006)	allelic to MFM2 (group 5)
Dilated cardiomyopathy, 1JJ	10.71	AD	CMD1JJ 615235	6q21	<i>LAMA4</i> 600133	Laminin, alpha-4	Knöll et al. (2007)	

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Dilated cardiomyopathy, 1KK	10.72	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	10.73	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	10.74	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	10.75	AD	CMD1NN 615916	3p25.2	<i>RAF1</i> 164760	V-Raf-1 murine leukemia viral oncogene homolog 1	Dhandapaney et al. (2014)	
Dilated cardiomyopathy related to <i>ILK</i>	10.76	AD		11p15.4	<i>ILK</i> 602366	Integrin-linked kinase	Knöll et al. (2007)	
Dilated cardiomyopathy related to <i>ANKRD1</i>	10.77	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	10.78	AR	CMD2A 611880	19q13.42	<i>TNNI3</i> 191044	Cardiac troponin I	Carballo et al. (2009)	allelic to CMH7, CMD1FF, RCM1 (group 10A)
Dilated cardiomyopathy 2B	10.79	AR	CMD2B 614672	7q21.2	<i>GATADI</i> 614518	GATA zinc finger domain-containing protein 1	Theis et al. (2011)	
Dilated cardiomyopathy, 3A	10.80	XR	CMD3A	Xq28	<i>TAZ</i> 300394	Tafazzin	Gedeon et al. (1995)	allelic to BTHS (group 10A)
Dilated cardiomyopathy, 3B	10.81	XR	CMD3B 302045	Xp21.2-p21.1	<i>DMD</i> 300377	Dystrophin	Muntoni et al. (1993) Milasin et al. (1996)	allelic to DMD (group 1)
Dilated cardiomyopathy related to nesprin-1	10.82	AD		6q25.2	<i>SYNE1</i> 608441	Spectrin repeat containing, nuclear envelope protein 1 (nesprin-1)	Puckelwartz et al. (2010)	allelic to EDMD4 (group 1) SCAR8 (group 13), AMCM (group 16)
Dilated cardiomyopathy related to MURC	10.83	AD		9q31.1	<i>CAVIN4</i> 617714	Caveolae-associated protein 4 (MURC)	Rodriguez et al. (2011)	
Dilated cardiomyopathy related to <i>DOLK</i>	10.84	AR	CDGM1 610768	9q34.11	<i>DOLK</i> 610746	Dolichol kinase	Kranz et al. (2007) Lefebvre et al. (2011)	
<b>Restrictive cardiomyopathies</b>								
Restrictive cardiomyopathy, 1	10.85	AD	RCM1 115210	19q13.42	<i>TNNI3</i> 191044	Cardiac troponin I	Mogensen et al. (2003)	allelic to CMH7, CMD1FF, CMD2A (group 10A)
Restrictive cardiomyopathy, 2	10.86		RCM2 609578	10q23.3	?	?	Zhang et al. (2005)	
Restrictive cardiomyopathy, 3	10.87	AD	RCM3 612422	1q32.1	<i>TNNT2</i> 191045	Cardiac Troponin T	Peddy et al. (2006)	allelic to CMH2, CMD1D and LVNC6 (group 10a)
Restrictive cardiomyopathy, 4	10.88	AD	RCM4 615248	10q21.3	<i>MYPN</i> 608517	Myopalladin	Purevjav et al. (2012)	allelic to NEM11 (group 3), CMH22 and CMD1KK (group 10A)
Restrictive cardiomyopathy, 5	10.89	AD	RCM5 617047	7q32.1	<i>FLNC</i> 102565	Filamin C	Brodehl et al. (2016)	allelic to MFM5 (group 5), MPD4 (group 4) and CMH26 (group 10A)
<b>Other non arrhythmic hereditary cardiomyopathies</b>								
Pompe disease, Glycogenosis, generalized, cardiac form (also listed in group 9)	10.90	AR	GSD2 232300	17q25.3	<i>GAA</i> 606800	Glucosidase, alpha; acid	Hers (1963) Martiniuk et al. (1990) Wokke et al. (1995)	Allelic to Pompe disease (formerly LGMD2V) (group 1)
Cardioskeletal myopathy with neutropenia and abnormal mitochondria (Barth syndrome)	10.91	XR	BTHS 302060	Xq28	<i>TAZ</i> 300394	Tafazzin	Bolhuis et al. (1991) Bione et al. (1996)	allelic to CMD3A (group 10A)
Left ventricular noncompaction, 1	10.92	AD	LVNC1 604169	18q12.1	<i>DTNA</i> 601239	Dystrobrevin, alpha	Ichida et al. (2001)	
Left ventricular noncompaction, 2	10.93		LVNC2 609470	11q15	?	?	Sasse-Klaassen et al. (2004)	
Left ventricular noncompaction, 3	10.94	AD	LVNC3 601493	10q23.2	<i>LDB3</i> 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	10.95	AD	LVNC4 613424	15q14	<i>ACTC1</i> 102540	Alpha-cardiac actin	Monserrat et al. (2007)	allelic to CMH11, CMD1R (group 10A)
Left ventricular noncompaction, 5	10.96	AD	LVNC5 613426	14q11.2	<i>MYH7</i> 160760	Myosin heavy chain 7 (beta), cardiac muscle	Klaassen et al. (2008)	allelic to CFTD, MSMA, MSMB and SPMM (group 3), MPD1 (group 4), CMH1 and CMD1S (group 10A)

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Left ventricular noncompaction, 6	10.97	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	10.98	AD	LVNC7 615092	18q11.2	<i>MIB1</i> 608677	Mindbomb, drosophila, homolog of	Luxan et al. (2013)	
Left ventricular noncompaction, 8	10.99	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	10.100	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	10.101	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)
Left ventricular hypertrabeculation/non compaction	10.102	AD		5q31	<i>MYOT</i> 604103	Myotilin	Finsterer et al. (2020)	Allelic to formerly LGMD1A (group 1), distal myopathy with myotilin defects (group 4), MFM3 and spheroid body myopathy (group 5)
Cardiovalvular dysplasia, X-linked (Myxomatous valvular dystrophy)	10.103	XR	CVD1 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 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	Wang et al. (1999) Shen et al. (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 3B, Fast-channel	11.6	AR	CMS3B 616322	2q37.1	<i>CHRNA1</i> 100690	Cholinergic receptor, nicotinic, alpha 1	Shen et al. (2003)	allelic to CMS1A (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	Shen et al. (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 4B, Fast-channel	11.7	AR	CMS4B 616324	17p13.2	<i>CHRNA1</i> 100690	Cholinergic receptor, nicotinic, alpha 1	Shen et al. (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	11.10	AR	CMS4C 608931	17p13.2	<i>CHRNA1</i> 100690	Cholinergic receptor, nicotinic, alpha 1	Shen et al. (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	11.8	AR	CMS2C 616314	17p13.1	<i>CHRNBI</i> 100710	Cholinergic receptor, nicotinic, beta 1	Quiram et al. (1999)	allelic to CMS2A (group 11)
Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	11.9	AR	CMS3C 616323	2q37.1	<i>CHRNA1</i> 100690	Cholinergic receptor, nicotinic, alpha 1	Shen et al. (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	11.10	AR	CMS4C 608931	17p13.2	<i>CHRNA1</i> 100690	Cholinergic receptor, nicotinic, alpha 1	Shen et al. (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 5	11.11	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	11.12	AR	CMS6 254210	10q11.23	<i>CHAT</i> 118490	Choline acetyltransferase	Ohno et al. (2001) Maselli et al. (2003)	
Myasthenic syndrome, congenital, 7, presynaptic	11.13	AD	CMS7 616040	1q32.1	<i>SYT2</i> 600104	Synaptotagmin 2	Herrmann et al. (2014)	Allelic to Distal motor neuropathy related to SYT2 (group 12)
Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	11.14	AR	CMS8 615120	1p36.33	<i>AGRN</i> 103320	Agrin	Huzé et al. (2009)	Allelic to FADS related to AGRN (group 16)
Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	11.15	AR	CMS9 616325	9q31.3	<i>MUSK</i> 601296	Muscle-specific receptor tyrosine kinase	Chevessier et al. (2004)	allelic to FADS (group 16)

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Myasthenic syndrome, congenital, 10	11.16	AR	CMS10 254300	4p16.3	<b>DOK7</b> 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	11.17	AR	CMS11 616326	11p11.2	<b>RAPSN</b> 601592	Receptor-associated protein of the synapse, 43kD (Rapsyn)	Ohno et al. (2002) Dunne et al. (2003)	allelic to FADS (group 16)
Myasthenia, congenital, 12, with tubular aggregates	11.18	AR	CMS12 610542	2p13.3	<b>GFPT1</b> 138292	Glutamine:fructose-6-phosphate amido transferase 1	Senderek et al. (2011)	
Myasthenic syndrome, congenital, 13, with tubular aggregates	11.19	AR	CMS13 614750	11q23.3	<b>DPAGTI</b> 191350	Dolichyl-phosphate N-acetylglucosamine phosphotransferase 1	Belaya et al. (2012)	
Myasthenic syndrome, congenital, 14, with tubular aggregates	11.20	AR	CMS14 616228	9q22.33	<b>ALG2</b> 607905	S. Cerevisiae homolog of ALG2 (alpha-1,3/1,6-mannosyl transferase)	Cossins et al. (2013)	
Myasthenic syndrome, congenital, 15, without tubular aggregates	11.21	AR	CMS15 607227	1p21.3	<b>ALG14</b> 612866	S. Cerevisiae homolog of ALG14 (UDP-N-acetylglucosaminyltransferase subunit)	Cossins et al. (2013)	
Myasthenic syndrome, congenital, 16	11.22	AR	CMS16 614198	17q23.3	<b>SCN4A</b> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Tsujino et al. (2003)	allelic to Congenital Myopathy related to <i>SCN4A</i> (group 3), HOKPP2, HYPF, PMC and K-aggravated myotonia (group 7), Severe foetal hypokinesia related to <i>SCN4A</i> (group 16)
Myasthenic syndrome, congenital, 17	11.23	AR	CMS17 616304	11p11.2	<b>LRP4</b> 604270	LDL receptor-related protein 4	Ohkawara et al. (2014)	
Myasthenic syndrome, congenital, 18	11.24	AD	CMS18 616330	20p12.2	<b>SNAP25</b> 600322	Synaptosomal associated protein 25	Shen et al. (2014)	
Myasthenic syndrome, congenital, 19	11.25	AR	CMS19 616720	10q22.1	<b>COL13A1</b> 120350	Collagen type XIII alpha 1 chain	Logan et al. (2015)	
Myasthenic syndrome, congenital, 20, presynaptic	11.26	AR	CMS20 617143	2q12.3	<b>SLC5A7</b> 608761	Solute carrier family 5 (choline transporter) member 7	Bauche et al. (2016)	allelic to HMN7A (group 12)
Myasthenic syndrome, congenital, 21, presynaptic	11.27	AR	CMS21 617239	10q11.23	<b>SLC18A3</b> 600336	Solute carrier family 18 (vesicular acetylcholine), member 3	O'Grady et al. (2016)	
Myasthenic syndrome, congenital, 22	11.28	AR	CMS22 616224	2p21	<b>PREPL</b> 609557	Prolyl endopeptidase-like	Regal et al. (2014)	
Presynaptic congenital myasthenic syndrome 23	11.29	AR	CMS23 618197	22q11.21	<b>SLC25A1</b> 190315	Solute carrier family 25 (mitochondrial carrier, citrate transporter), member 1	Chaouch et al. (2014)	
Presynaptic congenital myasthenic syndrome 24	11.30	AR	CMS24 618198	15q23	<b>MYO9A</b> 604875	Myosin IXA	O'Connor et al. (2016)	
Presynaptic congenital myasthenic syndrome 25	11.31	AR	CMS25 618323	12p13.31	<b>VAMP1</b> 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	11.32	AR	NPHS5 614199	3p21.31	<b>LAMB2</b> 150325	Laminin-beta 2	Maselli et al. (2009)	
Escobar syndrome (multiple pterygium syndrome)	11.33	AR	EVMP5 265000	2q37.1	<b>CHRNA3</b> 100730	Cholinergic receptor, nicotinic, gamma	Hoffman et al. (2006) Morgan et al. (2006)	
Myasthenic syndrome, with plectin defect	11.34	AR		8q24.3	<b>PLEC</b> 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 <i>GMPPB</i>	11.35	AR		3q21.31	<b>GMPPB</b> 615320	GDP-mannose pyrophosphorylase B	Belaya et al. (2015)	allelic to LGMDR19 (group 1); MEB/MDDGA14 and MDDGB14 (group 2)
Presynaptic congenital myasthenic syndrome	11.36	AR		20q13.33	<b>LAMA5</b> 601033	Laminin, alpha-5	Maselli et al. (2017)	
Presynaptic congenital myasthenic syndrome related to MUNC13-1	11.37	AR		19p13.11	<b>UNC13A</b> 609894	C. Elegans, homolog of UNC13A (MUNC13)	Engel et al. (2016)	
Congenital myasthenic syndrome related to RPH3A, presynaptic	11.38	AR		12q24.13	<b>RPH3A</b> 612159	Rabphilin 3A	Maselli et al. (2018)	

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<b>GROUP 12. SPINAL MUSCULAR ATROPHIES MOTONEURON DISEASES</b>								
<i>Spinal muscular atrophy related to SMN1</i>								
Spinal muscular atrophy, type I (Werdnig-Hoffman)	12.1	AR	SMA1 253300	5q13.2	<i>SMN1</i> 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	<i>SMN1</i> 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	<i>SMN1</i> 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	<i>SMN1</i> 600354	Survival of motor neuron 1	Brahe et al. (1995) Clermont et al. (1995)	allelic to SMA2, SMA3, SMA1 (group 12)
<i>Distal spinal muscular atrophy, recessive</i>								
Spinal muscular atrophy, distal, autosomal recessive 1 (with respiratory distress)	12.5	AR	DSMA1 604320	11q13.3	<i>IGHMBP2</i> 600502	Immunoglobulin Mu-binding protein 2	Grohmann et al. (1999, 2001)	allelic to CMT2S (group 14)
Spinal muscular atrophy, distal autosomal recessive 2	12.6	AR	DSMA2 605726	9p13.3	<i>SIGMARI</i> 601978	Sigma non-opioid intracellular receptor 1	Christodoulou et al. (2000) Li et al. (2015)	allelic to ALS16 (group 12)
Spinal muscular atrophy, distal autosomal recessive 3	12.7	AR	DSMA3 607088	11q13	?	?	Viollet et al. (2004)	
Spinal muscular atrophy, distal autosomal recessive 4	12.8	AR	DSMA4 611067	1p36.31	<i>PLEKHG5</i> 611101	Pleckstrin homology domain and RhoGEF domain-containing protein G5	Maystadt et al. (2006, 2007)	allelic to CMTRIC (group 14)
Spinal muscular atrophy, distal, autosomal recessive, 5	12.9	AR	DSMA5 614881	2q35	<i>DNAJB2</i> 604139	DnaJ/Hsp40 homolog, subfamily B, member 2	Blumen et al. (2012)	Allelic to CMT related to DNAJB2 (group 14)
Spinal muscular atrophy, distal, autosomal recessive	12.10	AR	DSMA	14q32.2	<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)
Spinal muscular atrophy with congenital bone fractures 1	12.11	AR	SMABF1 616866	15q22.31	<i>TRIP4</i> 604501	Thyroid hormone receptor interactor 4	Knierim et al. (2016)	allelic to MDCDC (group 2)
Spinal muscular atrophy with congenital bone fractures 2	12.12	AR	SMABF2 616867	10q22.1	<i>ASCC1</i> 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	12.13	AR	SMAPME 159950	8p22	<i>ASAH1</i> 613468	N-acylsphingosine amidohydrolase 1	Zhou et al. (2012)	allelic to FRBRL (#228000)
Spinal muscular atrophy and cerebellar hypoplasia	12.14	AR	PCH1C 616081	13q13.3	<i>EXOSC8</i> 606019	Exosome component 8	Boczonadi et al. (2014)	
Distal hereditary motor neuronopathy	12.15	AR		5q23.3	<i>HINT1</i> 601314	Histidine triad nucleotide-binding protein	Zhao et al. (2014)	Allelic to NMNAN (group 14)
<i>Distal spinal muscular atrophy, dominant</i>								
Neuronopathy, distal hereditary motor, type I	12.16	AD	HMN1 182960	7q34-q36	?	?	Gopinath et al. (2007)	
Neuronopathy, distal hereditary motor, type IIA	12.17	AD	HMN2A 158590	12q24.23	<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)
Neuronopathy, distal hereditary motor, type IIB	12.18	AD	HMN2B 608634	7q11.23	<i>HSPB1</i> 602195	Heat-shock 27-kD protein 1	Evgrafov et al. (2004)	allelic to CMT2F (group 14)
Neuronopathy, distal hereditary motor, type IIC	12.19	AD	HMN2C 613376	5q11.2	<i>HSPB3</i> 604624	Heat shock 27-kD protein 3	Kolb et al. (2010)	
Neuronopathy, distal hereditary motor, type IID	12.20	AD	HMN2D 615575	5q32	<i>FBXO38</i> 608533	F-box only protein 38	Sumner et al. (2013)	
Distal spinal muscular atrophy, distal with upper limb predominance (type V)	12.21	AD	HMN5A 600794	7p14.3	<i>GARS</i> 600287	Glycyl-tRNA synthetase	Christodoulou et al. (1995) Antonellis et al. (2003)	allelic to CMT2D (group 14)

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Distal spinal muscular atrophy type VA	12.22	AD	HMN5A 600794	11q12.3	<b>BSC12</b> 606158	Seipin	Windpassinger et al. (2004)	allelic to SPG17 (group 15)
Distal spinal muscular atrophy, type VB	12.23	AD	HMNS5B 614751	2p11.2	<b>REEP1</b> 609139	Receptor expression-enhancing protein 1	Beetz et al. (2012)	allelic to SPG31 (group 15)
Dominant distal hereditary motor neuropathy	12.24	AD	dHMN	16q22.1	<b>AARS1</b> 601065	Alanyl-tRNA synthetase 1	Zhao et al. (2012)	allelic to CMT2N (group 14)
Neuropathy, distal hereditary motor, type IX	12.25	AD	HMN9 61772	14q32.2	<b>WARS1</b> 191050	Tryptophanyl-tRNA synthetase 1	Tsai et al. (2017)	
Spinal muscular atrophy, distal, with vocal cord paralysis (Harper-Young)	12.26	AD	HMN7A 158580	2q12.3	<b>SLC5A7</b> 608761	Solute carrier family 5 (choline cotransporter), member 7	McEntagart et al. (2001) Barwick et al. (2012)	allelic to CMS20 (group 11)
Distal hereditary motor neuropathy type VIIIB	12.27	AD	HMN7B 607641	2p13.1	<b>DCTN1</b> 601143	Dynactin 1	Puls et al. (2003)	allelic to ALS related to DCTN1 (group 12)
Hereditary motor and sensory neuropathy V	12.28	AD	HMSN5 600361	4q34.3-q35.2	?	?	Muglia et al. (2008)	
Spinal muscular atrophy, distal, congenital non progressive	12.29	AD	HMN8 600175	12q24.11	<b>TRPV4</b> 605427	Transient receptor potential cation channel, subfamily V, member 4	van der Vleuten et al. (1998) Auer-Grumbach et al. (2010) Deng et al. (2010)	allelic to SPSMA (group 12), HSMN2C (group 14)
Scapuloperoneal spinal muscular atrophy	12.30	AD	SPSMA 181405	12q.24	<b>TRPV4</b> 605427	Transient receptor potential cation channel, subfamily V, member 4	Isozumi et al. (1996) Deng et al. (2010)	allelic to HMN8 (group 12), HSMN2C (group 14)
Spinal motor neuropathy	12.31			11q23.2	<b>RBM7</b> 612413	RNA-binding motif protein 7	Giunta et al. (2016)	
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 1	12.32	AD	SMALED1 158600	14q32.31	<b>DYNC1H1</b> 600112	Dynein, cytoplasmic 1, heavy chain 1	Harms et al. (2010, 2012)	allelic to CMT2O (group 14)
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 2A	12.33	AD	SMALED2A 615290	9q22.31	<b>BICD2</b> 609797	Bicaudal D homolog 2 (Drosophila)	Neveling et al. (2013) Oates et al. (2013) Peeters et al. (2013)	Allelic to SMALED2B (group 12) and Arthrogyrosis and BICD2-related neuromuscular disease (group 16)
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 2 B	12.34	AD	SMALED2B 618291	9q22.31	<b>BICD2</b> 609797	Bicaudal D homolog 2 (Drosophila)	Ravenscroft et al. (2016)	Allelic to SMALED2A (group 12) and Arthrogyrosis and BICD2-related neuromuscular disease (group 16)
Spinal muscular atrophy, late-onset, Finkel type	12.35	AD	SMAFK 182980	20q13.32	<b>VAPB</b> 605704	Vesicle-associated membrane protein-associated protein B	Nishimura et al. (2004)	allelic to ALS8 (group 12)
Spinal muscular atrophy, Jokela type	12.36	AD	SMAJ 615048	22q11.23	<b>CHCHD10</b> 615903	Coiled-coil-helix-coiled-coil-helix domain containing 10 (M)	Muller et al. (2014) Penttilä et al. (2012, 2015)	allelic to FTDALS2 (group 12), IMMD (group 16)
Distal motor neuropathy related to SYT2	12.37	AD		1q32.1	<b>SYT2</b> 600104	Synaptotagmin 2	Montes-Chinea et al. (2018)	Allelic to CMS7 (group 11)
Distal motor neuropathy	12.38	AD		9q34.11	<b>SPTANI</b> 182810	Spectrin, alpha, nonerythrocytic 1	Beijer et al. (2019)	
Distal hereditary motor neuropathies	12.39	AD		10q24.32	<b>GBF1</b> 603698	Golgi-specific brefeldin-A resistance factor 1	Mendoza-Ferreira et al. (2020)	
<b>Distal spinal muscular atrophy, X-linked</b>								
Spinal and bulbar muscular atrophy, X-linked, 1 (Kennedy disease)	12.40	XR	SBMX1 313200	Xq12	<b>AR</b> 313700	Androgen receptor	Fishbeck et al. (1986) La Spada et al. (1991)	
Spinal muscular atrophy, distal, X-linked 2	12.41	XR	SMAX2 301830	Xp11.3	<b>UBA1</b> 314370	Ubiquitin-like modifier-activating enzyme 1	Ramser et al. (2013) Diamini et al. (2013)	
Spinal muscular atrophy, distal, X-linked, 3	12.42	XR	SMAX3 300489	Xq21.1	<b>ATP7A</b> 300011	ATPase, Cu <sup>++</sup> transporting, alpha polypeptide	Takata et al. (2004) Kennerson et al. (2010)	
<b>Amyotrophic lateral sclerosis (ALS)</b>								
Amyotrophic lateral sclerosis 1 (dominant)	12.43	AD	ALS1 105400	21q22.11	<b>SOD1</b> 147450	Cu/Zn superoxide dismutase	Siddique et al. (1991, 1996) Rosen et al. (1993)	
Amyotrophic lateral sclerosis 1 (recessive)	12.44	AR	ALS1 105400	21q22.11	<b>SOD1</b> 147450	Cu/Zn superoxide dismutase	Andersen et al. (1995)	

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Amyotrophic lateral sclerosis 2, juvenile	12.45	AR	ALS2 205100	2q33.1	<i>ALS2</i> 606352	Alsin Rho guanine nucleotide exchange factor 2	Hentati et al. (1994) Yang et al. (2001) Hadano et al. (2001)	allelic to IAHSPL (group 15)
Amyotrophic lateral sclerosis 3	12.46	AD	ALS3 606640	18q21	?	?	Hand et al. (2002)	
Amyotrophic lateral sclerosis 4, juvenile	12.47	AD	ALS4 602433	9q34.13	<i>SETX</i> 608465	Senataxin	Chance et al. (1998) Chen et al. (2004) Moreira et al. (2004)	allelic to SCAR1 (group 13)
Amyotrophic lateral sclerosis 5, juvenile	12.48	AR	ALS5 602099	15q21.1	<i>SPG11</i> 610844	Spatacsin	Hentati et al. (1998) Orlacchio et al. (2010)	allelic to CMT2X (group 14)
Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	12.49	AD	ALS6 608030	16p11.2	<i>FUS</i> 137070	Fused in sarcoma	Sapp et al. (2003) Abalkhail et al. (2003) Kwiatkowski et al. (2009) Vance et al. (2009)	
Amyotrophic lateral sclerosis 7	12.50	AD	ALS7 608031	20p13	?	?	Sapp et al. (2003)	
Amyotrophic lateral sclerosis 8	12.51	AD	ALS8 608627	20q13.32	<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	12.52	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	12.53	AD	ALS10 612069	1p36.22	<i>TARDBP</i> 605078	TAR DNA-binding protein	Sreedharan et al. (2008)	
Amyotrophic lateral sclerosis 11	12.54	AD	ALS11 612577	6q21	<i>FIG4</i> 609390	FIG4, S. Cerevisiae, homolog of	Chow et al. (2009)	allelic to CMT4J (group 14)
Amyotrophic lateral sclerosis 12	12.55	AD, AR	ALS12 613435	10p13	<i>OPTN</i> 602432	Optineurin	Maruyama et al. (2010)	
Amyotrophic lateral sclerosis 13	12.56	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	12.57	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	12.58	XD	ALS15 300857	Xp11.21	<i>UBQLN2</i> 300264	Ubiquilin 2	Deng et al. (2011)	
Amyotrophic lateral sclerosis 16, juvenile	12.59	AR	ALS16 614373	9p13.3	<i>SIGMARI</i> 601978	Sigma non-opioid intracellular receptor 1	Al-Saif et al. (2011)	allelic to DSMA2 (group 12)
Amyotrophic lateral sclerosis 17	12.60	AD	ALS17 614696	3p11.2	<i>CHMP2B</i> 609512	Charged multivesicular body protein 2B	Parkinson et al. (2006) Cox et al. (20010)	
Amyotrophic lateral sclerosis 18	12.61	AD	ALS18 614808	17p13.2	<i>PFN1</i> 176610	Profilin 1	Wu et al. (2012)	
Amyotrophic lateral sclerosis 19	12.62	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	12.63	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	12.64	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	12.65	AD	ALS22 616208	2q35	<i>TUBA4A</i> 191110	Tubulin, alpha-4A	Smith et al. (2014)	
Amyotrophic lateral sclerosis 23	12.66	AD	ALS23 617839	10q22.3	<i>ANXA11</i> 602572	Annexin A11	Smith et al. (2017)	
Amyotrophic lateral sclerosis 24, susceptibility to	12.67	AD	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	12.68	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)

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Amyotrophic lateral sclerosis related to NEFH, susceptibility to	12.69	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	12.70	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	12.71	AD	ALS1 105400	2p13.1	<i>DCTN1</i> 601143	Dynactin 1	Munch et al. (2005)	allelic to HMN7B (group 12)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.72	AD	FTDALS1 105550	9p21.2	<i>C9orf72</i> 614260	Chromosome 9 open reading frame 72	Morita et al. (2006) DeJesus-Hernandez (2011)	
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.73	AD	FTDALS2 615911	22q11.23	<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)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.74	AD	FTDALS3 66437	5q35.3	<i>SQSTM1</i> 601530	Sequestosome 1	Fecto et al. (2011)	allelic to Welander-like syndrome and DMRV (group 4)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.75	AD	FTDALS4 616439	12q14.2	<i>TBK1</i> 604834	Tank-binding kinase 1	Cirulli et al. (2015) Freischmidt et al. (2015)	
<b>Others</b>								
Lethal Congenital Contracture Syndrome 1	12.76	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	12.77	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	12.78	AR	LCCS3 611359	19p13.3	<i>PIP5K1C</i> 606102	Phosphatidylinositol-4-phosphate 5-kinase, type I, gamma	Narkis et al. (2007)	
Lethal Congenital Contracture Syndrome 4	12.79	AR	LCCS4 614915	12q23.2	<i>MYBPC1</i> 160794	Myosin-binding protein C, slow type	Markus et al. (2012)	Allelic to DA1B (group 16)
Lethal Congenital Contracture Syndrome 5	12.80	AR	LCCS5 615368	19p13.2	<i>DNM2</i> 602378	Dynamin 2	Koutsopoulos et al. (2013)	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>CMTDIB</i> (group 14)
Lethal Congenital Contracture Syndrome 6	12.81	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	12.82	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	12.83	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	12.84	AR	LCCS9 616503	6q24.2	<i>ADGRG6</i> 612243	Adhesion G protein-coupled receptor G6	Ravenscroft et al. (2015)	
Lethal Congenital Contracture Syndrome 10	12.85	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	12.86	AR	LCCS11 617194	15q21.2	<i>GLDN</i> 608603	Gliomedin	Maluenda et al. (2016)	
Lethal Congenital Contracture Syndrome related to <i>TTN</i>	12.87	AR		2q31.2	<i>TTN</i> 188840	Titin	Chervinski et al. (2018)	Allelic to LGMDR10 (group 1), CNM related to <i>TTN</i> (group 3), MmD related to <i>TTN</i> (group 3), <i>SALMY</i> (group 3), <i>TMD</i> (group 4), <i>MFM9</i> (group 5), <i>CMH9</i> (group 10), <i>CMD1G</i> (group 10)
Spinal muscular atrophy with pontocerebellar hypoplasia, type 1b	12.88	AR	PCH1B 614678	9p13.2	<i>EXOSC3</i> 606489	Exosome component 3	Rudnik-Schöneborn et al. (2013)	
Spinal muscular atrophy with or without pontocerebellar hypoplasia	12.89	AR	PCH1 607596	14q32.2	<i>VRK1</i> 602168	Vaccinia related kinase 1	Renbaum et al. (2009) Stoll et al. (2016)	Allelic to DSMA (group 12), Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis (group 14)
Pontocerebellar hypoplasia with spinal muscular atrophy	12.90	AR	HSMN6B 616505	5q22.1	<i>SLC25A46</i> 610826	Solute Carrier Family 25, Member 46	van Dijk et al. (2017)	

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Brown-Vialetto-van Laere syndrome 1	12.91	AR	BVVL1 211530	20p13	<i>SLC52A3</i> 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	12.92	AR	BVVL2 614707	8q24.3	<i>SLC52A2</i> 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>	12.93	AR		5q13.3	<i>HEXB</i> 606873	Hexosaminidase B	Rattay et al. (2013)	allelic to Sandhoff disease, adult type (OMIM #268800)
Spinal muscular atrophy, related to <i>PRUNE1</i>	12.94	AR		1q21.3	<i>PRUNE1</i> 617413	Prune exopolyphosphatase 1	Iacomino et al. (2017)	
Spinal muscular atrophy with pontocerebellar hypoplasia related to <i>KIF26B</i>	12.95	AD		1q44	<i>KIF26B</i> 614026	Kinesin family member 26B	Wojcik et al. (2018)	
Peripheral neuropathy, myopathy, hoarseness and hearing loss	12.96	AD	PNMHH 614369	19q13.33	<i>MYH14</i> 608568	Myosin, heavy chain 14, non muscle	Choi et al. (2011)	
Lower motor neuron disease with respiratory failure related to <i>MAPT</i>	12.97	AD		17q21.31	<i>MAPT</i> 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	12.98	AR	PNRIID 618124	21q22.3	<i>MCM3AP</i> 603294	Minichromosome maintenance 3-associated protein	Schuurs-Hoeijmakers et al. (2013) Ylikallio et al. (2017)	

## 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	<i>PMP22</i> 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	<i>MPZ</i> 159440	Myelin protein zero	Bird et al. (1982) Guiloff et al. (1982) Hayasaka et al. (1993) Kulkens et al. (1993)	allelic to CMTDD, CMT2I, CMT2J, DSS and CHN (group14)
Charcot-Marie-Tooth disease, type 1C	14.3	AD	CMT1C 601098	16p13.13	<i>LITAF</i> 603795	Lipopolysaccharide-induced TNF-alpha factor	Street et al. (2002, 2003)	
Charcot-Marie-Tooth disease, type 1D	14.4	AD	CMT1D 607678	10q21.3	<i>EGR2</i> 129010	Early growth response 2 (Krox-20 homolog)	Warner et al. (1998) Street et al. (2003)	allelic to CHN and DSS (group 14)
Charcot-Marie-Tooth disease, type 1E, with deafness	14.5	AD	CMT1E 118300	17p12	<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 with congenital vertical talus	14.9	AD	CVT 192950	2q31.1	<i>HOXD10</i> 142984	Homeobox D10	Shrimpton et al. (2004)	
Slowed nerve conduction velocity	14.10	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	14.11	AD	HNARMID 608895	14q32.12	<i>FBLN5</i> 604580	Fibulin 5	Auer-Grumbach et al. (2011)	
Charcot-Marie-Tooth neuropathy	14.12	AD		6q21.31	<i>ITPR3</i> 147267	Inositol 1,4,5-triphosphate receptor, type 3	Rönkkö et al. (2020)	

## Dominant intermediate (CMTDI)

Charcot-Marie-Tooth disease, dominant intermediate A	14.13	AD	CMTDIA 606483	10q24.1-q25.1	?	?	Verhoeven et al. (2001)	
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Charcot-Marie-Tooth disease, dominant intermediate B	14.14	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), CNM1 (group 3), distal myopathy related to <i>DNM2</i> (group 4) and LCCS5 (group 12)
Charcot-Marie-Tooth disease, dominant intermediate C	14.15	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	14.16	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	14.17	AD	CMTDIE 614455	14q32.33	<i>INF2</i> 610982	Inverted formin 2	Boyer et al. (2011)	
Charcot-Marie-Tooth disease, dominant intermediate F	14.18	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	14.19	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	14.20	AD		1p21.2-p13.3	<i>C1orf194</i> 618682	Chromosome 1 open reading frame 194	Sun et al. (2019)	
<b>Autosomal recessive (AR-CMT1 or CMT4)</b>								
Charcot-Marie-Tooth disease, type 4A	14.21	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	14.22	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	14.23	AR	CMT4B2 604563	11p15.4	<i>SBF2</i> 607697	SET-binding factor 2	Azzedine et al. (2003)	
Charcot-Marie-Tooth disease, type 4B3	14.24	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	14.25	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	14.26	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	14.27	AR	CHN 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, 1	14.28	AR	CHN 605253	1q23.3	<i>MPZ</i> 159440	Myelin protein zero	Warner et al. (1996)	allelic to CMT1B, CMTDID, CMT2I, CMT2J, DSS (group 14)
Charcot-Marie-Tooth disease, type 4F	14.29	AR	CMT4F 614895	19q13.2	<i>PRX</i> 605725	Periaxin	Delague et al. (2000) Guilbot et al. (2001)	allelic to DSS (group14)
Neuropathy, hereditary motor and sensory, Russe type	14.30	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	14.31	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	14.32	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	14.33	AR	CMT4K 616684	9q34.2	<i>SURF1</i> 185620	Surfeit 1 (M)	Echaniz-Laguna et al. (2013)	
Charcot-Marie Tooth disease	14.34	AR		14q32	<i>AHNAK2</i> 608570	Ahnak nucleoprotein 2	Tey et al. (2019)	
Charcot-Marie Tooth disease related to <i>CNTNAP1</i>	14.35	AR		17q21.2	<i>CNTNAP1</i> 602346	Contactin-associated protein 1	Freed et al. (2019)	Allelic to LCCS7 (group 12)
<b>X-linked CMT1</b>								
Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	14.36	XD	CMTX1 302800	Xq13.1	<i>GJB1</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	14.37	XR	CMTX2 302801	Xp22.2	?	?	Ionasecu et al. (1992)	

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Charcot-Marie-Tooth neuropathy, X-linked recessive, 3	14.38	XR	CMTX3 302802	Xq26	<b>78kb Chr8 8q24.3 insertion</b>	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)	14.39	XR	COWCK 310490	Xq26.1	<b>AIFM1 300169</b>	Apoptosis-inducing factor, mitochondria-associated, 1 (M)	Priest et al. (1995) Rinaldi et al. (2012)	Allelic to COXPD6 (group 16)
Charcot-Marie-Tooth disease, X-linked recessive, 5	14.40	XR	CMTX5 311070	Xq22.3	<b>PRPS1 311850</b>	Phosphoribosyl pyrophosphate synthetase 1	Kim et al. (2007)	
Charcot-Marie-Tooth disease, X-linked dominant, 6	14.41	XD	CMTX6 300905	Xp22.11	<b>PDK3 300906</b>	Pyruvate dehydrogenase kinase, isoenzyme 3	Kennerson et al. (2013)	
<b>Déjerine-Sottas syndrome (DSS or CMT3)</b>								
Déjerine-Sottas hypertrophic neuropathy, dominant	14.42	AD	DSS 145900	17p12	<b>PMP22 601097</b>	Peripheral myelin protein 22	Roa et al. (1993)	allelic to CMT1A, CMT1E and HNPP (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.43	AD	DSS 145900	1q23.3	<b>MPZ 159440</b>	Myelin protein zero	Hayasaka et al. (1993)	allelic to CMT1B, CMTD1D, CMT2J, CMT2I and CHN (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.44	AD (digenic)	DSS 145900	10q21.3 and Xq13	<b>EGR2 129010 + GJBI 304040</b>	Early growth response 2 + Gap junction protein, beta 1	Chung et al. (2005)	<b>EGR2:</b> allelic to CMT1D and CHN (group 14)  <b>GJBI:</b> allelic to CMTX1 (group 14) allelic to CMT4F (group 14)
Déjerine-Sottas hypertrophic neuropathy, recessive	14.45	AR	DSS 145900	19q13.2	<b>PRX 605725</b>	Periaxin	Delague et al. (2000) Boerkoel et al. (2001)	
<b>B. Charcot-Marie-Tooth neuropathy, type 2 (axonal) = CMT2</b>								
<b>Autosomal dominant CMT2</b>								
Charcot-Marie-Tooth disease, type 2A1	14.46	AD	CMT2A1 118210	1p36.22	<b>KIF1B 605995</b>	Kinesin family member 1B	Zhao et al. (2001)	
Charcot-Marie-Tooth disease, axonal, type 2A2A	14.47	AD	CMT2A2A 609260	1p36.22	<b>MFN2 608507</b>	Mitofusin 2	Ben Othmane et al. (1993) Züchner et al. (2004)	allelic to CMT2A2B and HMSN6A (group14)
Charcot-Marie-Tooth disease, type 2B	14.48	AD	CMT2B 600882	3q21.3	<b>RAB7 602298</b>	RAS-associated protein RAB7	Kwon et al. (1995) Pericak-Vance et al. (1997) Kok et al. (2003)	
Hereditary motor and sensory neuropathy, type IIC	14.49	AD	HMSN2C 606071	12q24.11	<b>TRPV4 605427</b>	Transient receptor potential cation channel, subfamily V, member 4	Klein et al. (2003) McEntagart et al. (2005) Auer-Grumbach et al. (2010) Deng et al. (2010) Landouere et al. (2010)	allelic to HMN8 and SPSMA (group 12)
Charcot-Marie-Tooth disease, type 2D	14.50	AD	CMT2D 601472	7p14.3	<b>GARS 600287</b>	Glycyl-tRNA synthetase	Ionasecu et al. (1996) Antonellis et al. (2003)	allelic to HMN5A (group12)
Charcot-Marie-Tooth disease, type 2E	14.51	AD	CMT2E 607684	8p21.2	<b>NEFL 162280</b>	Neurofilament, light polypeptide	Birouk et al. (2003) Claramunt et al. (2005)	allelic to CMT1F, CMTD1G (group 14)
Charcot-Marie-Tooth disease, type 2F	14.52	AD	CMT2F 606595	7q11.23	<b>HSPB1 602195</b>	Heat-shock 27-kD protein 1	Ismailov et al. (2001) Evgrafov et al. (2004)	allelic to HMN2B (group 14)
Charcot-Marie-Tooth disease, type 2H	14.53	AD	CMT2H 607731	8q13-q23	?	?	Barhoumi et al. (2001)	maybe allelic to CMT4A (group14)
Charcot-Marie-Tooth disease, type 2I	14.54	AD	CMT2I 607677	1q23.3	<b>MPZ 159440</b>	Myelin protein zero	Auer-Grumbach et al. (2003)	allelic to CMT1B, CMTD1D, CMT2J, DSS, CHN (group 14)
Charcot-Marie-Tooth disease, type 2J	14.55	AD	CMT2J 607736	1q23.3	<b>MPZ 159440</b>	Myelin protein zero	De Jonghe et al. (1999) Chapon et al. (1999)	allelic to CMT1B, CMTD1D, CMT2I, DSS, CHN (group 14)
Charcot-Marie-Tooth disease, type 2K	14.56	AD, AR	CMT2K 607831	8q21.11	<b>GDAP1 606598</b>	Ganglioside-induced differentiation-associated protein 1	Nelis et al. (2002) Birouk et al. (2003) Claramunt et al. (2005)	allelic to CMT4A and CMTRIA (group 14)
Charcot-Marie-Tooth disease, type 2L	14.57	AD	CMT2L 608673	12q24.23	<b>HSPB8 608014</b>	Heat-shock 22-kD protein 8	Tang et al. (2004, 2005)	allelic to Rimmed vacuole myopathy and distal myopathy and motor neuropathy (group 4) and HMN2A (group 12)
Charcot-Marie-Tooth disease, type 2N	14.58	AD	CMT2N 613287	16q22.1	<b>AARS1 601065</b>	Alanyl-tRNA synthetase 1	Latour et al. (2010)	Allelic to dHMN (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)
Charcot-Marie-Tooth disease, type 2O	14.59	AD	CMT2O 614228	14q32.31	<i>DYNCH1H</i> 600112	Dynein, cytoplasmic 1, heavy chain 1	Weedon et al. (2011)	allelic to SMALED1 (group 12)
Charcot-Marie-Tooth disease, type 2P	14.60	AD	CMT2P 614436	9q33.3-q34.1	<i>LRSAMI</i> 610933	Leucine rich repeat and sterile alpha motif containing 1	Nelis et al. (2004) Guernsey et al. (2010) Weterman et al. (2012)	
Charcot-Marie-Tooth disease, type 2Q	14.61	AD	CMT2Q 615025	10p14	<i>DHTKDI</i> 614984	Dehydrogenase E1 and transketolase domain containing 1	Xu et al. (2012)	
Charcot-Marie-Tooth disease, type 2U	14.62	AD	CMT2U 616280	12q13.3	<i>MARS</i> 156560	Methionyl-tRNA synthetase	Gonzalez et al. (2013)	
Charcot-Marie-Tooth disease, type 2V	14.63	AD	CMT2V 616491	17q21.2	<i>NAGLU</i> 609701	N-acetyl-alpha-glucosaminidase	Tetreault et al. (2015)	
Charcot-Marie-Tooth disease, type 2W	14.64	AD	CMT2W 616625	5q31.3	<i>HARS</i> 142810	Histidyl-tRNA synthetase	Vester et al. (2013) Safka-Brozokova et al. (2015)	
Charcot-Marie-Tooth disease, type 2Y	14.65	AD	CMT2Y 616687	9p13.3	<i>VCP</i> 601023	Valosin-containing protein	Gonzalez et al. (2014)	allelic to Scapuloperoneal myopathy (group 1), IBMPPFD1 (groups 4 and 5), ALS14 (group 12)
Charcot-Marie-Tooth disease, type 2Z	14.66	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	14.67	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	14.68	AD	CMT2DD 618036	1p13.1	<i>ATPIA1</i> 182310	ATPase, Na <sup>+</sup> /K <sup>+</sup> transporting, alpha-1 polypeptide	Lassuthova et al. (2018)	
Hereditary motor and sensory neuropathy, Okinawa type	14.69	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	14.70	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>	14.71	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	14.72	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>	14.73	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>	14.74	AD	CMT2	20p12.2	<i>JAG1</i> 601920	Jagged 1	Sullivan et al. (2020)	
<b>Autosomal recessive CMT2</b>								
Charcot-Marie-Tooth disease, axonal, type 2A2B	14.75	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	14.76	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, formerly LGMD1B (group 1), MDCL (group 2), CMD1A (group 10A) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370] Allelic to AOA4 (group 14)
Charcot-Marie-Tooth disease, axonal, type 2B2	14.77	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	14.78	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>	14.79	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	14.80	AR	NMAN 137200	5q23.3	<i>HINT1</i> 601314	Histidine triad nucleotide binding protein 1	Zimon et al. (2012)	Allelic to Distal hereditary motor neuropathy (group 12)
Charcot-Marie-Tooth disease, recessive intermediate, A	14.81	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)

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Charcot-Marie-Tooth disease, recessive intermediate, B	14.82	AR	CMTRIB 613641	16q23.1	<i>KARS</i> 601421	Lysyl-tRNA synthetase	McLaughlin et al. (2010)	
Charcot-Marie-Tooth disease, recessive intermediate, C	14.83	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	14.84	AR	CMTRID 616039	12q24.31	<i>COX6A1</i> 602072	Cytochrome c oxidase subunit 6a1 (M)	Tamiya et al. (2014)	
Charcot-Marie-Tooth disease, type 2R	14.85	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	14.86	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)
Charcot-Marie-Tooth disease, type 2T	14.87	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	14.88	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	14.89	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	14.90	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>	14.91	AR		22q13.33	<i>SCO2</i> 604272	Cytochrome c oxidase assembly protein 2	Rebello et al. (2018)	allelic to CEMCOX1 (group 10A)
Charcot-Marie-Tooth disease, axonal; related to <i>SACS</i>	14.92	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	14.93	AR	SORDD 618912	15q21.1	<i>SORD</i> 182500	Sorbitol Dehydrogenase	Cortese et al. (2020)	

**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	14.94	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)	
Hereditary sensory and autonomic neuropathy, type IB with cough and gastroesophageal reflux	14.95	AD	HSAN1B 608088	3p24-p22	?	?	Kok et al. (2004)	
Hereditary sensory and autonomic neuropathy, type IC	14.96	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	14.97	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 neuropathy, type IIB	14.98	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	14.99	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	14.100	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	14.101	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	14.102	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	14.103	AR	HSAN6 614653	6p12.1	<i>DST</i> 113810	Dystonin	Edvardson et al. (2012)	
Neuropathy, hereditary sensory and autonomic, type VII	14.104	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	14.105	AR	HSAN8 616488	9q34.12	<i>PRDM12</i> 616458	PR Domain-containing protein 12	Chen et al. (2015)	
Hereditary sensory neuropathy, type ID	14.106	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	14.107	AD	HSN1E 614116	19p13.2	<i>DNMT1</i> 126375	DNA methyltransferase 1	Klein et al. (2011)	
Neuropathy, hereditary sensory, type IF	14.108	AD	HSN1F 615632	11q13.1	<i>ATL3</i> 609369	Atlantin GTPase 3	Kornak et al. (2014)	
Hereditary sensory neuropathy, type IIC	14.109	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)	14.110	AR	AXPC1 609033	1q32.3	<i>FLVCRI</i> 609144	Feline leukemia subgroup C receptor 1	Rajadhyaksha et al. (2010)	
Absence of pain, Congenital	14.111	AR		22q11.21	<i>CLTCL1</i> 601273	Clathrin, heavy polypeptide-like 1	Nahorski et al. (2015)	
Marsili syndrome (insensitivity to pain, congenital, AD)	14.112	AD	MARSIS 147430	14q11.2	<i>ZFHX2</i> 617828	Zinc finger homeobox 2	Habib et al. (2018)	
Erythromelalgia, Primary	14.113	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	14.114	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	14.115	AR		1q25.3	<i>NMNAT2</i> 608701	Nicotinamide nucleoside adenyltransferase 2	Huppke et al. (2019)	
<b>E. Other complex neuropathy syndromes</b>								
Peripheral neuropathy and agenesis of the corpus callosum (Charlevoix disease)	14.116	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	14.117	AD		1p34.3	<i>GJB3</i> 603324	Gap junction protein, beta 3	Lopez-Bigas et al. (2001)	
Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy)	14.118	AD	HNA 162100	17q25.3	<i>SEPT9</i> 604061	Septin 9	Pellegrino et al. (1996) Kuhlenbaumer et al. (2005)	
Giant axonal neuropathy-1	14.119	AR	GAN1 256850	16q23.2	<i>GANI</i> 605379	Gigaxonin	Ben Hamida et al. (1997) Bomont et al. (2000) Klein et al. (2014)	
Giant axonal neuropathy-2	14.120	AD	GAN2 610100	1q23.2	<i>DCAF8</i> 615820	DDB1- and CUL4-associated factor 8		
Congenital cataracts, facial dysmorphism and neuropathy	14.121	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	14.122	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	14.123	AR	256840	5p15.2	<i>CCT5</i> 610150	Chaperonin containing T-complex polypeptide1, subunit 5	Bouhouche et al. (2006)	
Neuronal intranuclear inclusion diseases	14.124	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)	
Hereditary peripheral neuropathy (CMT ?)	14.125	AD		17q25.1	<i>SLC9A3RI</i> ( <i>EBP50</i> ) 604990	Solute carrier family 9, member 3, regulator 1	Song et al. (2020)	

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<b>Group 16. OTHER NEUROMUSCULAR DISORDERS</b>								
Torsion dystonia 1, early onset	16.1	AD	DYT1 128100	9q34.11	<b>TORIA</b> 605204	Torsin 1A	Ozelius et al. (1997) Ikeuchi et al. (1999)	
Myoclonic dystonia 11	16.2	AD	DYT11 159900	7q21.3	<b>SGCE</b> 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	<b>ELP1</b> 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	<b>TTR</b> 176300	Transthyretin	Costa et al. (1978) Tawara et al. (1983) Saraiva et al. (1995)	
Fibrosis of extraocular muscles, congenital, 1	16.5	AD	CFEOM1 CFEOM3B 135700	12q12	<b>KIF21A</b> 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	<b>PHOX2A</b> 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	<b>TUBB3</b> 602661	Tubulin, beta-3	Doherty et al. (1999) Tischfield et al. (2010)	
Fibrosis of extraocular muscles, congenital, 5	16.8	AR	CFEOM5 66219	4q25	<b>COL25A1</b> 610004	Collagen, type XXV, alpha-1	Shinwari et al. (2015)	
Arthrogryposis, distal, type 1A	16.9	AD	DA1A 108120	9p13.3	<b>TPM2</b> 190990	Tropomyosin-2 (beta)	Sung et al. (2003)	allelic to NEM4 and CAPM2 (group 3), DA2B (group 16)
Arthrogryposis, distal, type 1B	16.10	AD	DA1B 614335	12q23.2	<b>MYBPC1</b> 160794	Myosin-binding proteinC, slow type	Gurnett et al. (2010)	Allelic to LCCS4 (group 12)
Arthrogryposis, distal, type 2A, Freeman-Sheldon syndrome	16.11	AD	DA2A 193700	17p13.1	<b>MYH3</b> 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	<b>TNNI2</b> 191043	Troponin I, fast skeletal	Sung et al. (2003a) Kimber et al. (2006)	
Arthrogryposis, distal, type 2B2	16.13	AD	DA2B2 618435	11p15.5	<b>TNNT3</b> 600692	Troponin T3, fast skeletal	Sung et al. (2003)	
Arthrogryposis, distal, type 2B3	16.14	AD	DA2B3 618436	17p13.1	<b>MYH3</b> 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	<b>TPM2</b> 190990	Tropomyosin-2 (beta)	Tajsharghi et al. (2007) Ochala et al. (2007)	allelic NEM4 and CAPM2 (group 3), DA1A (group 16)
Arthrogryposis, distal, type 3	16.16	AD	DA3 114300	18p11.22-p11.21	<b>PIEZO2</b> 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	<b>PIEZO2</b> 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	<b>ECELI</b> 605896	Endothelin-converting enzyme like 1	McMillin et al. (2013)	
Arthrogryposis and Distal, type 7 (Trismus-pseudocamptodactyly syndrome)	16.19	AD	DA7 158300	17p13.1	<b>MYH8</b> 160741	Myosin heavy chain, 8, skeletal muscle, perinatal	Veugelers et al. (2004) Toydemir et al. (2006b)	
Arthrogryposis, distal, type 10	16.20	AD	DA10 187370	2q31.3-q32.1	?	?	Stevenson et al. (2006a) Stevenson et al. (2006b)	
Arthrogryposis multiplex congenita with nesprin-1 defect	16.21	AR	AMCM 618484	6q25.2	<b>SYNE1</b> 608441	Spectrin repeat containing, nuclear envelope 1 (Nesprin-1)	Attali et al. (2009)	allelic to EDMD4 (group 1), DCM related to SYNE1 (group 10A) and SCAR8 (group 13)
Arthrogryposis and BICD2-related neuromuscular disease	16.22	AD		9q22.31	<b>BICD2</b> 609797	Bicaudal D homolog 2 (Drosophila)	Storbeck et al. (2017)	Allelic to SMALED2A and SMALED2B (group 12)
Arthrogryposis and muscular dysplasia	16.23	AD		7q31.2	<b>MET</b> 164860	MET protooncogene	Zhou et al. (2019)	
Arthrogryposis related to ASCCI	16.24	AR		10q22.1	<b>ASCCI</b> 614215	Activating signal cointegrator 1 complex subunit 1	Böhm et al. (2019)	allelic to SMABF2 (group 12)

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Neurodevelopmental disorder with microcephaly, arthrogryposis and structural brain anomalies	16.25	AR	NEDMABA 618622	2q21.1	<b>SMPD4</b> 610457	Sphingomyelin phosphodiesterase 4, neutral membrane	Magini et al. (2019)	
Fetal akinesia deformation sequence 1	16.26	AR	FADS1 208150	9q31.3	<b>MUSK</b> 601296	Muscle-specific receptor tyrosine kinase	Tan-Sindhunata et al. (2015)	allelic to CMS9 (group 11)
Fetal akinesia deformation sequence 3	16.27	AR	FADS3 618389	4p16.3	<b>DOK7</b> 610285	Downstream of tyrosin kinase 7	Vogt et al. (2009)	allelic to CMS10 (group 11)
Fetal akinesia deformation sequence 2	16.28	AR	FADS2 618388	11p11.2	<b>RAPSN</b> 601592	Receptor-associated protein of the synapse, 43kD (Rapsyn)	Vogt et al. (2008)	allelic to CMS11 (group 11)
Fetal akinesia deformation sequence 4	16.29	AR	FADS4 618393	17p13.2	<b>NUP88</b> 602552	Nucleoporin 88kD	Bonnin et al. (2018)	
Fetal akinesia deformation sequence related to <i>RYR1</i>	16.30	AR		19q13.2	<b>RYR1</b> 180901	Ryanodine receptor	Romero et al. (2003) McKie et al. (2014) Alkhunaizi et al. (2019)	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), MHS1 (group 8)
Fetal akinesia deformation related to <i>AGRN</i>	16.31	AR		1p36.33	<b>AGRN</b> 103320	Agrin	Geremek et al. (2020)	Allelic to CMS8 (group 11)
Severe foetal hypokinesia related to <i>SCN4A</i>	16.32	AR		17q23.3	<b>SCN4A</b> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Zaharieva et al. (2019)	allelic to Congenital Myopathy related to <i>SCN4A</i> (group 3), HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1	16.33	AD	PEOA1 157640	15q26.1	<b>POLG</b> 174763	Polymerase, DNA, gamma (M)	Van Goethem et al. (2001)	allelic to SANDO (group 13), MTDPS4A, MTDPS4B, PEOB1 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2	16.34	AD	PEOA2 609283	4q35	<b>SLC25A4</b> 103220	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 (M)	Kaukonen et al. (2000)	Allelic to MTDPS12A and MTDPS12B (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3	16.35	AD	PEOA3 609286	10q24.31	<b>TWINK</b> 606075	Twinkle, mtDNA helicase (M)	Suomalinen et al. (1997) Spelbrink et al. (2001)	allelic to IOSCA (group 13)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4	16.36	AD	PEOA4 610131	17q23.3	<b>POLG2</b> 604983	Polymerase DNA, gamma 2 (M)	Longley et al. (2006)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5	16.37	AD	PEOA5 613077	8q22.3	<b>RRM2B</b> 604712	Ribonucleotide reductase, M2 B (M)	Tyynismaa et al. (2009)	allelic to MTDPS8A (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6	16.38	AD	PEOA6 615156	10q21.3	<b>DNA2</b> 601810	DNA replication helicase 2 (M)	Ronchi et al. (2013)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 1	16.39	AR	PEOB1 258450	15q26.1	<b>POLG</b> 174763	Polymerase, DNA, gamma (M)	Deschauer et al. (2007)	allelic to SANDO (group 13), MTDPS4A, MTDPS4B, PEOA1 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	16.40	AR	PEOB2 616479	2p25.3	<b>RNASEH1</b> 604123	Ribonuclease H1 (M)	Reyes et al. (2015)	

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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)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3	16.41	AR	PEOB3 617069	16q21	<i>TK2</i> 188250	Thymidine kinase, mitochondrial (M)	Tyynismaa et al. (2012)	allelic to MTDPS2 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4	16.42	AR	PEOB4 617070	2p13.1	<i>DGUOK</i> 601465	Deoxyguanosine kinase (M)	Ronchi et al. (2012)	Allelic to MTDPS3 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5	16.43	AR	PEOB5 618098	17p11.2	<i>TOP3A</i> 601243	DNA topoisomerase III	Nicholls et al. (2018)	
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	16.44	AR	MTDPS1 603041	22q13.33	<i>TYMP</i> 131222	Thymidine phosphorylase	Nishino et al. (1999)	
Mitochondrial DNA depletion syndrome 2 (myopathic type)	16.45	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)	16.46	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)	16.47	AR	MTDPS4A 203700	15q26.1	<i>POLG</i> 174763	Polymerase, DNA, gamma (M)	Naviaux and Nguyen (2004)	Allelic to SANDO (group 13), MTDPS4B, PEOA1 and PEOB1 (group 16)
Mitochondrial DNA depletion syndrome 4B (MNGIE type)	16.48	AR	MTDPS4B 613662	15q26.1	<i>POLG</i> 174763	Polymerase, DNA, gamma (M)	Van Goethem et al. (2003)	Allelic to SANDO (group 13), MTDPS4A, PEOA1 and PEOB1 (group 16)
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	16.49	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)	16.50	AR	MTDP8A 612075	8q22.3	<i>RRM2B</i> 604712	Ribonucleotide reductase, M2B (M)	Bourdon et al. (2007)	allelic to PEOA5 and MTDPS8B (group 16)
Mitochondrial DNA depletion syndrome 8B (MNGIE type)	16.51	AR	MTDP8B 612075	8q22.3	<i>RRM2B</i> 604712	Ribonucleotide reductase, M2B (M)	Bourdon et al. (2007)	allelic to PEOA5 and MTDPS8A (group 16)
Mitochondrial DNA depletion syndrome 9 (Encephalomyopathic type with methylmalonic aciduria)	16.52	AR	MTDPS9 245400	2p11.2	<i>SUCLG1</i> 611224	Succinate-CoA ligase, alpha subunit (M)	Ostergaard et al. (2007)	
Mitochondrial DNA depletion syndrome 11	16.53	AR	MTDPS11 615084	20p11.23	<i>MGME1</i> 615076	Mitochondrial genome maintenance exonuclease 1 (M)	Kornblum et al. (2013)	
Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type)	16.54	AD	MTDPS12A 617184	4q35.1	<i>SLC25A4</i> 103220	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 (M)	Thompson et al. (2016)	Allelic to PEOA2 and MTDPS12B (group 16)
Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type)	16.55	AR	MTDPS12B 615418	4q35.1	<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)	16.56	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)	16.57	AR	MTDPS14 616896	3q29	<i>OPA1</i> 605290	OPA1 protein (M)	Spiegel et al. (2016)	Allelic to Progressive external ophthalmoplegia with optic atrophy, optic atrophy 1 with deafness (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)
Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression	16.58	AR	MECREN 618416	19p13.11	<b>SLC25A42</b> 610823	Solute carrier family 25, member 42 (M)	Shamseldin et al. (2015)	
Progressive external ophthalmoplegia with optic atrophy, optic atrophy 1 with deafness	16.59	AD	125250	3q29	<b>OPA1</b> 605290	OPA1 protein (M)	Amati-Bonneau et al. (2008) Hudson et al. (2008)	Allelic to MTDPS14 (group 16)
Myopathy, lactic acidosis, and sideroblastic anemia 1	16.60	AR	MLASA1 600462	12q24.33	<b>PUS1</b> 608109	Pseudourine synthase 1 (M)	Bykhovskaya et al. (2004) Fernandez-Vizarra (2007)	
Myopathy, lactic acidosis, and sideroblastic anemia-2	16.61	AR	MLASA2 613561	12p11.21	<b>YARS2</b> 610957	Tyrosyl-tRNA synthetase 2 (M)	Riley et al. (2010)	Allelic to CMTDIC (group 14)
Isolated mitochondrial myopathy	16.62	AD	IMMD 616209	22q11.23	<b>CHCHD10</b> 615903	Coiled-coil-helix-coiled-coil-helix domain containing 10 (M)	Ajrroud-Driss et al. (2014)	allelic to FTDALS2 and SMAJ (group 12)
Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis	16.63	AD	POIKTMP 615704	11q12.1	<b>FAM111B</b> 615584	Family with sequence similarity 111, member B	Mercier et al. (2015)	
Combined oxidative phosphorylation deficiency 6;	16.64	XL	COXPD6 300816	Xq26.1	<b>AIFM1</b> 300169	Apoptosis-inducing factor, mitochondria-associated, 1 (M)	Morton et al. (2017)	Allelic to COCK (group 14)
Mitochondrial myopathy with severe neurological manifestations	16.65	AR		8q24.13	<b>TMEM65</b> 616609	Transmembrane protein 65	Nasli et al. (2017)	
Mitochondrial complex IV deficiency	16.66	AR	220110	2q33.3	<b>FASTKD2</b> 612322	Fast kinase domains 2 (M)	Yoo et al. (2017)	
Mitochondrial complex IV deficiency	16.67	AR	220110	16p11.2	<b>COX6A2</b> 6020009	Cytochrome c-oxidase, subunit 6A2 (M)	Inoue et al. (2019)	
Infantile-onset multisystem disease with progressive muscle weakness	16.68	AR	IMNEPD 616263	17q23.1	<b>PTRH2</b> 608625	Peptidyl-tRNA hydrolase 2	Hu et al. (2014)	
Dyskinetic cerebral palsy, partial agenesis of the corpus callosum and mitochondrial myopathy	16.69	AR		3p25.1	<b>MRPS25</b> 611987	Mitochondrial ribosomal protein S25 (M)	Bugiardini et al. (2019)	
Myopathy, mitochondrial and cerebellar ataxia (also listed in group 13)	16.70	AR	MMYAT 617675	1q22	<b>MSTO1</b> 617619	Misato 1, Mitochondrial Distribution and Morphology Regulator (M)	Nasca et al. (2017) Ardicli et al. (2019) Donkervoort et al. (2019)	allelic to CMD related to <i>MSTO1</i> (group 2)
Early onset of mitochondrial myopathy	16.71	AR		17q13.3	<b>TIMM22</b> 607251	Translocase of inner mitochondrial membrane 22 (M)	Pacheu-Grau et al. (2018)	
Coenzyme Q10 deficiency 1	16.72	AR	COQ10D1 607426	4q21.23	<b>COQ2</b> 609825	Coenzyme Q2 (M)	Quinzii et al. (2006)	
Autosomal recessive spinocerebellar ataxia, 9 with ubiquinone deficiency (also listed in group 13)	16.73	AR	SCAR9 (COQ10D4) 612016	1q42.13	<b>ADCK3</b> ( <i>COQ8A</i> , <i>CABC1</i> ) 606980	Coenzyme Q8A (M)	Lagier-Tourenne et al. (2008) Mollet et al. (2008)	
Coenzyme Q10 deficiency 5	16.74	AR	COQ10D5 614654	16q21	<b>COQ9</b> 612837	Coenzyme Q9 (M)	Rahman et al. (2001) Duncan et al. (2009)	
Coenzyme Q10 deficiency 6	16.75	AR	COQ10D6 614650	14q24.3	<b>COQ6</b> 614647	Coenzyme Q6 (M)	Heeringa et al. (2011)	
Coenzyme Q10 deficiency 7	16.76	AR	COQ10D7 607426	9q34.11	<b>COQ4</b> 612898	Coenzyme Q4 (M)	Brea-Calvo et al. (2015)	
Coenzyme Q10 deficiency 8	16.77	AR	COQ10D8 616733	16p12.3	<b>COQ7</b> 601683	Coenzyme Q7 (M)	Freyer et al. (2015)	
Sensory motor axonal neuropathy and myopathy	16.78	AR		19p13.2	<b>FDX2</b> 614585	Ferredoxin (M)	Gurgel-Giannetti et al. (2019)	
Spectraplaklinopathy	16.79	AR		1p34.3	<b>MACF1</b> 608271	Microtubule-actin cross-linking factor 1	Kang et al. (2020)	allelic to LIS9 (#618325)



**NEW REFERENCES****GROUP 1. MUSCULAR DYSTROPHIES**

Vissing J, Johnson K, Topf A, Nafissi S, Diaz-Manera J, French VM et al. *POPDC3* gene variants associated with a new form of limb girdle muscular dystrophy. *Ann. Neurol.* 86: 832–843, 2019. PMID: 31610034 [Item #1.52]

Di Costanzo S, Balasubramanian A, Pond HL, Rozkalne A, Pantaleoni C, Saredi S, et al. *POMK* mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. *Hum. Molec. Genet.* 23: 5781–5792, 2014. PMID: 24925318 [Item #1.61]

**GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES**

Villar-Quiles RN, Gomez-Garcia de la Banda M, Barois A, Bouchet-Séraphin C, Romero NB, Rio M, et al. Muscular, Ocular and Brain Involvement Associated with a De Novo 11q13.2q14.1 Duplication: Contribution to the Differential Diagnosis of Muscle-Eye-Brain Congenital Muscular Dystrophy. *J Neuromuscul Dis.* 2020;7(1):69–76. PMID: 31796684 [Item #2.31]

van Tol W, Ashikov A, Korsch E, Abu Bakar N, Willemsen MA, Thiel C, et al. A mutation in mannose-phosphate-dolichol utilization defect 1 reveals clinical symptoms of congenital disorders of glycosylation type I and dystroglycanopathy. *JIMD Rep.* 2019;50(1):31–39. PMID: 31741824 [Item #2.36]

**GROUP 3. CONGENITAL MYOPATHIES**

Pegoraro E, Gavassini BF, Borsato C, Melacini P, Vianello A, Stramere, R, et al. *MYH7* gene mutation in myosin storage myopathy and scapulo-peroneal myopathy. *Neuromusc. Disord.* 17: 321–329, 2007. PMID: 17336526 [Item #3.62]

Dafsari HS, Kocaturk NM, Daimagüler HS, Brunn A, Dötsch J, Weis J, et al. Bi-allelic mutations in uncoordinated mutant number-45 myosin chaperone B are a cause for congenital myopathy. *Acta Neuropathol Commun.* 2019;7(1):211. PMID: 31852522 [Item #3.63]

Garibaldi M, Rendu J, Brocard J, Lacene E, Fauré J, Brochier G, et al. ‘Dusty core disease’ (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. *Acta Neuropathol Commun.* 2019 Jan 5;7(1):3. PMID: 30611313 [Item #3.64]

**GROUP 4. DISTAL MYOPATHIES**

Lee Y, Jonson PH, Sarparanta J, Palmio J, Sarkar M, Vihola A, et al. *TIA1* variant drives myodegeneration in multisystem proteinopathy with *SQSTM1* mutations. *J Clin Invest.* 2018;128(3):1164–1177. PMID: 29457785 [Item #4.8]

Kiiski KJ, Lehtokari VL, Vihola AK, Laitila JM, Huovinen S, Sagath LJ, et al. Dominantly inherited distal nemaline/cap

myopathy caused by a large deletion in the nebulin gene. *Neuromuscul Disord.* 2019;29(2):97–107. PMID: 30679003 [Item #4.12]

Palmio J, Jonson PH, Inoue M, Sarparanta J, Bengoechea R, Savarese M, et al. Mutations in the J domain of *DNAJB6* cause dominant distal myopathy. *Neuromuscul Disord.* 2020;30(1):38–46. PMID: 31955980 [Item #4.23]

Savarese M, Palmio J, Poza JJ, Weinberg J, Olive M, Cobo AM, et al. Actininopathy: a new muscular dystrophy caused by *ACTN2* dominant mutations. *Ann. Neurol.* 85: 899–906, 2019. PMID: 30900782 [Item #4.25]

Ghaoui R, Palmio J, Brewer J, Lek M, Needham M, Evilä A, et al. Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy [published correction appears in *Neurology.* 2016 Mar 15;86(11):1077]. *Neurology.* 2016;86(4):391–398. PMID: 26718575 [Item #4.26]

**GROUP 5. OTHER MYOPATHIES**

Hedberg-Oldfors C, Meyer R, Nolte K, Abdul Rahim Y, Lindberg C, Karason K, et al. Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. *Brain* 143: 2406–2420, 2020. PMID: 32779703 [Item #5.11]

Deng J, Yu J, Li P, Luan X, Cao L, Zhao J, et al. Expansion of GGC repeat in *GIPCI* is associated with oculopharyngodistal myopathy. *Am. J. Hum. Genet.* 106: 793–804, 2020. PMID: 32413282 [Item #5.20]

**GROUP 10A. HEREDITARY CARDIOMYOPATHIES NON ARRHYTHMOGENIC**

Finsterer J, Stöllberger C, Hasun M, Riedhammer K, Wagner M. Multisystem Myotilinopathy, including Myopathy and Left Ventricular Noncompaction, due to the *MYOT* Variant c.179C>T. *Case Rep Cardiol.* 2020;2020:5128069. PMID: 32509353 [Item #10.102]

**GROUP 12. SPINAL MUSCULAR ATROPHIES, MOTOR NEURON DISEASES**

Mendoza-Ferreira N, Karakaya M, Cengiz N, Beijer D, Brigatti KW, Gonzaga-Jauregui C, et al. De Novo and Inherited Variants in *GBF1* are Associated with Axonal Neuropathy Caused by Golgi Fragmentation. *Am J Hum Genet.* 2020 Oct 1;107(4):763–777. PMID: 32937143 [Item #12.39]

Markus B, Narkis G, Landau D, Birk RZ, Cohen I, Birk OS. Autosomal recessive lethal congenital contractural syndrome type 4 (LCCS4) caused by a mutation in *MYBPC1*. *Hum. Mutat.* 33: 1435–1438, 2012. PMID: 22610851 [Item #12.79]

Koutsopoulos OS, Kretz C, Weller CM, Roux A, Mojzisova H, Böhm J, et al. Dynamin 2 homozygous mutation in humans with a lethal congenital syndrome. *Europ. J. Hum. Genet.* 21: 637–642, 2013. PMID: 23092955 [Item #12.80]

Patel N, Smith LL, Faqeih E, Mohamed J, Gupta, VA, Alkuraya, FS. *ZBTB42* mutation defines a novel lethal congenital contracture syndrome (LCCS6). *Hum. Molec. Genet.* 23: 6584–6593, 2014. PMID: 25055871, [Item #12.81]

Lakhani S, Doan R, Almureikhi M, Partlow JN, Al Saffar M, Elsaid MF, et al. Identification of a novel *CNTNAP1* mutation causing arthrogryposis multiplex congenita with cerebellar and cerebellar atrophy. *Europ. J. Med. Genet.* 60: 245–249, 2017. PMID: 28254648 [Item #12.82]

Agolini E, Cherchi C, Bellacchio E, Martinelli D, Cocciafero D, Cutrera R, et al. Expanding the clinical and molecular spectrum of lethal congenital contracture syndrome 8 associated with biallelic variants of *ADCY6*. *Clin Genet.* 2020;97(4):649–654. PMID: 31846058 [Item #12.83]

Ravenscroft G, Nolent F, Rajagopalan S, Meireles AM, Paavola KJ, Gaillard D, et al. Mutations of *GPR126* are responsible for severe arthrogryposis multiplex congenita. *Am. J. Hum. Genet.* 96: 955–961, 2015. PMID: 26004201 [Item #12.84]

Casey JP, Brennan K, Scheidel N, McGettigan P, Lavin PT, Carter S, et al. Recessive *NEK9* mutation causes a lethal skeletal dysplasia with evidence of cell cycle and ciliary defects. *Hum. Molec. Genet.* 25: 1824–1835, 2016. PMID: 26908619 [Item #12.85]

Maluenda J, Manso C, Quevarec L, Vivanti A, Marguet F, Gonzales M, et al. Mutations in *GLDN*, encoding gliomedin, a critical component of the nodes of Ranvier, are responsible for lethal arthrogryposis. *Am. J. Hum. Genet.* 99: 928–933, 2016. PMID: 27616481 [Item #12.86]

Chervinsky E, Khayat M, Soltsman S, Habiballa H, Elpeleg O, Shalev S. A homozygous *TTN* gene variant associated with lethal congenital contracture syndrome. *Am J Med Genet A.* 2018;176(4):1001–1005. PMID: 29575618 [Item #12.87]

Schuurs-Hoeijmakers JHM, Vulto-van Silfhout AT, Vissers LELM, van de Vondervoort IIGM, van Bon, BWM, de Ligt, J, et al. Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. *J. Med. Genet.* 50: 802–811, 2013. PMID: 24123876 [Item #12.98]

Ylikallio E, Woldegebriel R, Tumiati M, Isohanni P, Ryan MM, Stark Z, et al. *MCM3AP* in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. *Brain* 140: 2093–2103, 2017. PMID: 28633435 [Item #12.98]

#### GROUP 14. HEREDITARY MOTOR SENSORY NEUROPATHIES (HMSN)

Rönkkö J, Molchanova S, Revah-Politi A, Pereira EM, Auranen M, Toppila J, et al. Dominant mutations in *ITPR3* cause Charcot-Marie-Tooth disease. *Ann Clin Transl Neurol.* 2020 Oct;7(10):1962–1972. PMID: 32949214 [Item #14.12]

Freed AS, Weiss MD, Malouf EA, Hisama FM. *CNTNAP1* mutations in an adult with Charcot Marie Tooth disease.

*Muscle Nerve.* 2019;60(5):E28–E30. PMID: 31397905 [Item #14.35]

Sullivan JM, Motley WW, Johnson JO, Aisenberg WH, Marshall KL, Barwick K, et al. Dominant mutations of the Notch ligand *Jagged1* cause peripheral neuropathy. *J Clin Invest.* 2020;130(3):1506–1512. PMID: 32065591 [Item #14.74]

Cortese A, Zhu Y, Rebelo AP, Negri S, Courel S, Abreu L, et al. Biallelic mutations in *SORD* cause a common and potentially treatable hereditary neuropathy with implications for diabetes. *Nature Genet.* 52: 473–481, 2020. Note: Erratum: *Nature Genet.* 52: 640 only, 2020. PMID: 32367058 [Item #14.93]

Michiels JJ, te Morsche RHM, Jansen JBMJ, Drenth JPH. Autosomal dominant erythralgia associated with a novel mutation in the voltage-gated sodium channel alpha-subunit *Na(v)1.7*. *Arch. Neurol.* 62: 1587–1590, 2005. PMID: 16216943 [Item #14.113]

Faber CG, Hoeijmakers JGJ, Ahn H-S, Cheng X, Han C, Choi J.-S, et al. Gain of function *Na(v)1.7* mutations in idiopathic small fiber neuropathy. *Ann. Neurol.* 71: 26–39, 2012. PMID: 21698661 [Item #14.113]

Zhang XY, Wen J, Yang W, Wang C, Gao L, Zheng LH, et al. Gain-of-function mutations in *SCN11A* cause familial episodic pain. *Am. J. Hum. Genet.* 93: 957–966, 2013. PMID: 24207120 [Item #14.114]

Huang J, Han C, Estacion M, Vasylyev D, Hoeijmakers JG, Gerrits MM, et al. Gain-of-function mutations in sodium channel *Na(v)1.9* in painful neuropathy. *Brain* 137: 1627–1642, 2014. PMID 24776970 [Item #14.114]

Huppke P, Wegener E, Gilley J, Angeletti C, Kurth I, Drenth JPH, et al. Homozygous *NMNAT2* mutation in sisters with polyneuropathy and erythromelalgia. *Exp Neurol.* 2019 Oct;320:112958. PMID: 31132363 [Item #14.115]

Song GJ, Gupta DP, Rahman MH, Park HT, Al Ghoulleh I, Bisello A, et al. Loss-of-function of *EBP50* is a new cause of hereditary peripheral neuropathy: *EBP50* functions in peripheral nerve system. *Glia.* 2020;68(9): 1794–1809. PMID: 32077526 [Item #14.125]

#### GROUP 16. OTHER NEUROMUSCULAR DISORDERS

Magini P, Smits DJ, Vandervore L, Schot R, Columbaro M, Kasteleijn E, et al. Loss of *SMPD4* causes a developmental disorder characterized by microcephaly and congenital arthrogryposis. *Am. J. Hum. Genet.* 105: 689–705, 2019. PMID: 31495489 [Item #16.25]

Geremek M, Dudarewicz L, Obersztyn E, Paczkowska M, Smyk M, Sobocka K, et al. Null variants in *AGRN* cause lethal fetal akinesia deformation sequence. *Clin Genet.* 2020;97(4):634–638. PMID: 31730230 [Item #16.31]

Kang L, Liu Y, Jin Y, Li M, Song J, Zhang Y, et al. Mutations of *MACF1*, Encoding Microtubule-Actin Crosslinking-Factor 1, Cause Spectraplaklinopathy. *Front Neurol.* 2020 Jan 15;10:1335. PMID: 32010038 [Item #16.79]