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# The 2025 version of the gene table of neuromuscular disorders (nuclear genome)

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## General features

This table used to be published annually in the December issue since 2010, but is now published in the January issue. Its purpose is to provide the reader of *Neuromuscular Disorders* with an updated list of monogenic neuromuscular diseases due to a primary defect residing in the nuclear genome. It comprises diseases in which the causative gene is known or at least localized on a chromosome, if not yet identified. Diseases for which the locus has not been mapped or which are due to defects involving mitochondrial genes are not included.<sup>1</sup>

The diseases are classified into 17 groups, with this year an additional group devoted to mitochondrial myopathies (group 16):

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. Mitochondrial myopathies.
17. 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 <https://www.musclegenetable.fr>. It is designed to cope with the complexity described above. In this version the data are cross-referenced and linked

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<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.)

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

### Overview of the new data in the 2025 printed version of the gene table (in this issue)

There are 21 new items, marked by background shading. Altogether they comprise **15 additional genes**, **5 additional phenotypic variants** caused by a gene already listed in the 2024 version. One locus has been identified.

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

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

### Citation of the gene table

- Printed version: Benarroch L, Bonne G, Rivier F, Procaccio V, Hamroun D. The 2025 version of the gene table of neuromuscular disorders. *Neuromuscul Disord.* xx (x), xx–xxx.
- Online version: Gene Table of Neuromuscular Disorders: <https://www.musclegenetable.fr>

### Contact

Users of the gene table are kindly requested to send any comments on the printed and/or the online version to [gisele.bonne@inserm.fr](mailto:gisele.bonne@inserm.fr).

### Acknowledgements

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*over the maintenance of the “Muscle Gene Table” he initiated in 1991. We sincerely wish him an enjoyable retirement from the Gene Table, knowing he will keep a kindly eye on it. We sincerely thank for their careful review of entries: Tanya Stojkovic for groups 12 and 14.*

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### New in the 2025 printed version of the gene table

#### 15 genes added:

*SNUPN* (item # 1.42)  
*HMGCS1* (item # 2.18)  
*JPH1* (item # 3.66)  
*SOX8* (item # 3.67)  
*ABCD3* (item # 5.27)  
*NUTM2B-AS1* (item # 5.28)  
*ABCC9* (item # 5.48)  
*PACSIN3* (item # 5.49)  
*VEZF1* (item # 10.78)  
*CAP2* (item # 10.89)  
*FLII* (item # 10.90)  
*PURA* (item # 11.45)  
*COX18* (item # 12.48)  
*CIAO1* (item # 16.37)  
*THOC2* (item # 17.26)

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

*TARDBP* (item # 4.30)  
*RTN2* (item # 12.15)  
*TIA1* (item # 12.79)  
*NARS1* (item # 14.79)  
*TECPR2* (item # 14.114)

#### 1 locus identified

*HNRNPA1* (item # 4.7)

#### 23 new key references

## Gene table of monogenic neuromuscular disorders (nuclear genome only) Vol. 46, January 2025

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	XLR	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) Hodgson et al (1986) Romeo et al (1988) Bione et al (1994, 1995) Klauck et al (1995) Nigro et al (1995) Bonne et al (1999) Worman and Bonne (2007)	allelic to CMD3B (group10A)
Emery-Dreifuss muscular dystrophy, X-linked, type 1	1.2	XLR	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) Bonne et al (1999) Worman and Bonne (2007)	
Emery-Dreifuss muscular dystrophy 2, autosomal dominant	1.3	AD	EDMD2 181350	1q22	<i>LMNA</i> 150330	Lamin A/C	Bonne et al (1999) Worman and Bonne (2007)	Allelic to EDMD3 (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	1.4	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), 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	1.5	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	1.6	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 6, X-linked	1.7	XLR	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 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-Kurekci et al (2014) Fichtman et al (2019)	Allelic to CMS (group 11)
Facio-scapulo-humeral muscular dystrophy, type 1	1.10	AD	FSHD1 158900	4q35	<i>DUX4*</i> 606009 (*inappropriate reactivation)	Double homeobox 4	Wijmenga et al (1990-1993) Upadhyaya et al. (1990, 1992) Wright et al (1993) van Deutekom et al (1993) Gabellini et al (2002) Van der Maarel et al (2005) Gabellini et al (2006) Petrov et al (2006) Lemmers et al (2010)	

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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)
Facio-scapulo-humeral muscular dystrophy, type 2	1.11	AD	FSHD2 158901	18p11.32	<b>SMCHD1</b> * 614982 (*causing inappropriate reactivation of DUX4* 606009)	Structural maintenance of chromosomes flexible hinge domain containing 1	de Greef et al (2010) Sacconi et al (2012) Lemmers et al (2012) Sacconi et al (2013)	allelic to Bosma Arhinia Microphthalmia Syndrome; BAMS (#603457)
Facio-scapulo-humeral muscular dystrophy	1.12	AR	FSHD3 619477	1p13.3	<b>LRF1</b> 615354	Ligand-Dependent Nuclear Receptor-Interacting Factor 1	Hamanaka et al (2020)	
Lipodystrophy, congenital generalized, type 4	1.13	AD	CGL4 613327	17q21.2	<b>CAVIN1</b> 603198	Caveolae-associated protein 1	Hayashi et al (2009)	
Muscular dystrophy with gnathodiaphyseal dysplasia	1.14	AR		11p14.3	<b>ANOS</b> ( <b>TMEM16E</b> ) 608662	Anoctamin 5	Shaibani et al (2021)	allelic to LGMDR12 (group 1) and MMD3 (group 4)
Muscular dystrophy with hearing loss and ovarian insufficiency syndrome	1.15	AR	MDHLO 619518	1q42.3	<b>GGPS1</b> 606982	Geranylgeranyl Diphosphate Synthase 1	Foley et al (2020)	
<b>Limb girdle muscular dystrophies, dominant</b>								
Muscular dystrophy, limb-girdle, autosomal dominant 1 (formerly LGDM1E)	1.16	AD	LGMD1E (LGMD1E) 603511	7q36.3	<b>DNAJB6</b> 611332	Hsp40 homologue, subfamily B, number 6	Speer et al (1999), Sarparanta et al (2012) Harms et al (2012)	allelic to Rimmed vacuole distal myopathy and distal myopathy (group 4)
Muscular dystrophy, limb-girdle, autosomal dominant 2 (Formerly LGMD1F)	1.17	AD	LGMD2E (LGMD1F) 608423	7q32.1	<b>TNPO3</b> 610032	Transportin 3	Palenzuela et al (2003) Melià et al (2013) Torella et al (2013)	allelic to Congenital Myopathy related to TNPO3 (group 3)
Muscular dystrophy, limb-girdle, autosomal dominant 3 (Formerly LGMD1G)	1.18	AD	LGMD3 (LGMD1G) 609115	4q21.22	<b>HNRNPDL</b> 607137	Heterogeneous nuclear ribonucleoprotein D-like	Starling et al (2005) Vieira et al (2014)	
Muscular dystrophy, limb-girdle, autosomal dominant 4	1.19	AD	LGMD4 (LGMD1I) 618129	15q15.1	<b>CAPN3</b> 114240	Calpain-3	Vissing et al (2016) Martinez-Thompson et al (2018)	allelic to LGMDR1 (group 1)
Rippling muscle disease 2 (Formerly LGMD1C)	1.20	AD	RMD2 606072 (LGMD1C 607801)	3p25.3	<b>CAV3</b> 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)
Muscular dystrophy, limb-girdle, type 1H	1.21	AD	LGMD1H 613530	3p25.1-p23	?	?	Bisceglia et al (2010)	
LGMD related to <i>KBTBD13</i>	1.22	AD		15q22.31	<b>KBTBD13</b> 613727	Kelch repeat and BTB/POZ domain containing protein 13	Garibaldi et al. (2018)	allelic to NEM6 (group 3)
LGMD related to <i>CACNA1S</i> , late-onset	1.23	AD		1q32.1	<b>CACNA1S</b> 114208	Calcium channel, voltage-dependent, L type, alpha 1S subunit	López-Hernández et al (2021)	allelic to Congenital myopathy with ophthalmoplegia related to CACNA1S, CMYO18 (group 3), HOKPP1 (group 7) and MHS5 (group 8)
<b>Limb girdle muscular dystrophies, recessive</b>								
Muscular dystrophy, limb-girdle, autosomal recessive 1 (Formerly LGMD2A)	1.24	AR	LGMDR1 (LGMD2A) 253600	15q15.1	<b>CAPN3</b> 114240	Calpain-3	Beckmann et al. (1991) Young et al. (1992) Richard et al (1995, 1997)	allelic to LGMD4 (group 1)
Muscular dystrophy, limb-girdle, autosomal recessive 2 (Formerly LGMD2B)	1.25	AR	LGMDR2 (LGMD2B) 253601	2p13.2	<b>DYSF</b> 603009	Dysferlin	Bashir et al (1994) Bashir et al (1998) Liu et al (1998)	allelic to MMD1 (group 4)
Muscular dystrophy, limb-girdle, autosomal recessive 3 (Formerly LGMD2D)	1.26	AR	LGMDR3 (LGMD2D) 608099	17q21.33	<b>SGCA</b> 600119	Alpha-sarcoglycan	Roberds et al (1994) Piccolo et al (1995) Passos-Bueno et al (1995) Ljunggren et al (1995) Carrié et al (1997)	
Muscular dystrophy, limb-girdle, autosomal recessive 4 (Formerly LGMD2E)	1.27	AR	LGMDR4 (LGMD2E) 604286	4q12	<b>SGCB</b> 600900	Beta-sarcoglycan	Lim et al (1995) Bönnemann et al (1995) Bönnemann et al (1996)	

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Muscular dystrophy, limb-girdle, autosomal recessive 5 (Formerly LGMD2C)	1.28	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)	
Muscular dystrophy, limb-girdle, autosomal recessive 6 (Formerly LGMD2F)	1.29	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)
Muscular dystrophy, limb-girdle, autosomal recessive 7 (Formerly LGMD2G)	1.30	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)
Muscular dystrophy, limb-girdle, autosomal recessive 8 (Formerly LGMD2H)	1.31	AR	LGMDR8 (LGMD2H) 254110	9q33.1	<i>TRIM32</i> 602290	Tripartite motif-containing 32	Weiler et al. (1998) Frosk et al. (2002)	allelic to sarcotubular myopathy (group 3)
Muscular dystrophy, limb-girdle, autosomal recessive 10 (Formerly LGMD2J)	1.32	AR	LGMDR10 (LGMD2J) 608807	2q31.2	<i>TTN</i> 188840	Titin	Hackman et al. (2003)	allelic to CNM related to <i>TTN</i> , MmD related to <i>TTN</i> and <i>CMYO5</i> (group 3), <i>TMD</i> (group 4), <i>MFM9</i> (group 5), <i>CMH9</i> (group 10), <i>CMD1G</i> (group 10), <i>LCCS</i> related to <i>TTN</i> (group 12)
Muscular dystrophy, limb-girdle, autosomal recessive 12 (Formerly LGMD2L)	1.33	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 Muscular dystrophy with gnathodiaphyseal dysplasia (group 1) and <i>MMD3</i> (group 4)
Muscular dystrophy, limb-girdle, autosomal recessive 17 (Formerly LGMD2Q)	1.34	AR	LGMDR17 (LGMD2Q) 613723	8q24.3	<i>PLEC</i> 601282	Plectin	Gundesli et al. (2010)	allelic to LGMD with ophtalmoplegia (group 1), <i>EBSMD</i> (group 5), and Myasthenic syndrome related to <i>PLEC</i> (group 11)
Muscular dystrophy, limb-girdle, autosomal recessive 18 (Formerly LGMD2S)	1.35	AR	LGMDR18 (LGMD2S) 615356	4q35.1	<i>TRAPPC11</i> 614138	Trafficking protein particle complex 11	Bögershausen et al. (2013)	allelic to CMD related to <i>TRAPPC11</i> (group 2)
Muscular dystrophy, limb-girdle, autosomal recessive 21 (Formerly LGMD2Z)	1.36	AR	LGMDR21 (LGMD2Z) 617232	3q13.33	<i>POGLUT1</i> 615618	Protein O-Glucosyltransferase 1	Servian-Morilla et al. (2016)	
Muscular dystrophy, limb-girdle, autosomal recessive 23	1.37	AR	LGMDR23 618138	6q22.33	<i>LAMA2</i> 156225	Laminin 2, Heavy chain (laminin alpha2 chain of merosin)	Gavassini et al. (2011)	allelic to <i>MDC1A</i> (group 2)
Muscular dystrophy, limb-girdle, autosomal recessive 25 (Formerly LGMD2X)	1.38	AR	LGMDR25 (LGMD2X) 616812	6q21	<i>BVES</i> (= <i>POPDC1</i> ) 604577	Blood vessel epicardial substance	Schindler et al. (2016)	
Muscular dystrophy, limb-girdle, autosomal recessive 26	1.39	AR	LGMDR26 618848	6q21	<i>POPDC3</i> 605824	Popeye domain-containing protein 3	Vissing et al. (2019)	
Muscular dystrophy, limb-girdle, autosomal recessive 27	1.40	AR	LGMDR27 619566	14q32.33	<i>JAG2</i> 602570	Jagged 2	Coppens et al. (2021)	
Muscular dystrophy; limb-girdle, autosomal recessive 28	1.41	AR	LGMDR28 620375	5q13.3	<i>HMGCR</i> 142910	3-Hydroxy-3-methylglutaryl-CoA reductase	Yogev et al. (2023) Morales-Rosado et al. (2023)	
Muscular dystrophy, limb-girdle, autosomal recessive 29	1.42	AR	LGMDR29 620793	15q24.2	<i>SNUPN</i> 607902	Surpentin 1	Iruzubieta et al. (2024)	
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C1 Formerly LGMDR11 (LGMD2K)	1.43	AR	MDDGC1 (LGMDR11) 609308	9q34.13	<i>POMT1</i> 607423	Protein O-mannosyltransferase 1	Balci et al. (2005) D'Amico et al. (2006)	Allelic to <i>MDDGA1</i> (group 2) and <i>MDDGB1</i> (group 2)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C2 Formerly LGMDR14 (LGMD2N)	1.44	AR	MDDGC2 (LGMDR14) 613158	14q24.3	<i>POMT2</i> 607439	Protein O-mannosyl transferase 2	Biancheri et al. (2007)	Allelic to <i>MDDGA2</i> and <i>MDDGB2</i> (group 2)

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [M])	Key references	Other allelic disease(s) (group in this table)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C3 Formerly LGMDR15 (LGMD2O)	1.45	AR	MDDGC3 (LGMDR15) 613157	1p34.1	<b>POMGNT1</b> 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 and MDDGB3 (group 2)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C4 Formerly LGMDR13 (LGMD2M)	1.46	AR	MDDGC4 (LGMDR13) 611588	9q31.2	<b>FKTN</b> 607440	Fukutin	Murakami et al (2006) Godfrey et al (2006)	Allelic to MDDGA4 and MDDGB4 (group 2), CMD1X (group 10A)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C5 Formerly LGMDR9 (LGMD2I)	1.47	AR	MDDGC5 (LGMDR9) 607155	19q13.32	<b>FKRP</b> 606596	Fukutin related protein	Driss et al (2000) Brockington et al (2001a)	Allelic to MDDGA5 and MDDGB5 (group 2),
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C7 Formerly LGMDR20 (LGMD2U)	1.48	AR	MDDGC7 (LGMDR20) 616052	7p21.2-p21.1	<b>CRPPA</b> 614631	Isoprenoid synthase domain containing protein	Tasca et al (2013)	Allelic to MDDGA7 (group 2)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C8 Formerly LGMDR24	1.49	AR	MDDGC8 (LGMDR24) 618135	3p22.1	<b>POMGNT2</b> 614828	Protein O-mannose beta-1,4-N-acetylglucosaminyl transferase 2	Endo et al (2015)	allelic to MDDGA8 (group 2)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C9 Formerly LGMDR16 (LGMD2P)	1.50	AR	MDDGC9 (LGMDR16) 613818	3p21.31	<b>DAG1</b> 128239	Dystrophin-associated glycoprotein 1 (alpha-dystroglycan)	Hara et al (2011)	Allelic to MDDGA9 (group 2)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C12	1.51	AR	MDDGC12 616094	8p11.21	<b>POMK</b> 615247	Protein-O-mannose kinase	Di Costanzo et al. (2014)	Allelic to MDDGA12 (group 2)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C14 Formerly LGMDR19 (LGMD2T)	1.52	AR	MDDGC14 (LGMDR19) 615352	3p21.31	<b>GMPFB</b> 615320	GDP-mannose pyrophosphorylase B	Carss et al (2013) Cabrera-Serrano et al (2015)	Allelic to MDDGA14, MDDGB14 (group 2) and congenital myasthenic syndrome (group 11)
Muscular dystrophy-dystroglycanopathy (Limb-girdle), type C15	1.53	AR	MDDGC15 612937	1q22	<b>DPM3</b> 605951	Dolichyl-phosphate mannosyltransferase polypeptide 3	Lefeber et al (2009)	Allelic to MDDGB15 (group 2)
Pompe disease (Formerly LGMD2V)	1.54	AR	GSD2 232300	17q25.3	<b>GAA</b> 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	<b>LIMS2</b> (= <i>PINCH2</i> ) 607908	LIM and senescent cell antigen-like domains 2	Chardon et al (2015)	
Scapuloperoneal muscular dystrophy and dropped head syndrome	1.56	AD, AR	600416	9p13.3	<b>VCP</b> 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	1.57	AR		8q24.3	<b>PLEC</b> 601282	Plectin	Fattahi et al (2015)	allelic to LGMDR17 (group 1), EBSMD (group 5), myasthenic syndrome related to <i>PLEC</i> (group 11)
LGMD related to <i>PYROXD1</i>	1.58	AR		12p12.1	<b>PYROXD1</b> 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)

## GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES

Congenital muscular dystrophy with merosin deficiency	2.1	AR	MDC1A 607855	6q22.33	<b>LAMA2</b> 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	<b>COL6A1</b> 120220	Collagen type VI subunit alpha 1	Jöbsis et al (1996)	allelic to UCMD1 (group 2)
Bethlem myopathy 1	2.3	AD	BTHLM1 158810	21q22.3	<b>COL6A2</b> 120240	Collagen type VI subunit alpha 2	Jöbsis et al (1996)	allelic to UCMD1 and Congenital myosclerosis (group 2)
Bethlem myopathy 1	2.4	AD	BTHLM1 158810	2q37.3	<b>COL6A3</b> 120250	Collagen type VI subunit alpha 3	Speer et al (1996) Bertini et al (1998) Pan et al (1998)	allelic to UCMD1 (group 2)
Bethlem myopathy 1	2.5	AR	BTHLM1 158810	21q22.3	<b>COL6A2</b> 120240	Collagen type VI subunit alpha 2	Gualandi et al (2009)	allelic to UCMD1 and Congenital myosclerosis (group 2)

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Ullrich congenital muscular dystrophy 1	2.6	AR	UCMD1 254090	21q22.3	<b>COL6A1</b> 120220	Collagen, type VI, subunit alpha 1	Pan et al (2003) Giusti et al (2005)	allelic to BTHLM1 (group 2)
Ullrich congenital muscular dystrophy 1	2.7	AR	UCMD1 254090	21q22.3	<b>COL6A2</b> 120240	Collagen, type VI, subunit alpha 2	Vanegas et al (2001) Higuchi et al (2001)	allelic to BTHLM1 and congenital myosclerosis (group 2)
Ullrich congenital muscular dystrophy 1	2.8	AR	UCMD1 254090	2q37.3	<b>COL6A3</b> 120250	Collagen type VI subunit alpha 3	Demir et al (2002)	allelic to BTHLM1 (group 2)
Ullrich congenital muscular dystrophy 2	2.9	AR	UCMD2 616470	6q13-q14	<b>COL12A1</b> 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	<b>COL12A1</b> 120320	Collagen type XII alpha 1 chain	Zou et al (2014)	Allelic to UCMD2 (group 2), CMD related to COL12A1 (group 2)
COL12A1-related congenital muscular dystrophy	2.11	AD		6q13-q14	<b>COL12A1</b> 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	<b>COL6A2</b> 120240	Collagen type VI subunit alpha 2	Merlini et al (2008)	allelic to UCMD1 and BTHLM1 (group 2)
Congenital myopathy 3 with rigid spine	2.13	AR	CMYO3 602771	1p36.11	<b>SELENON</b> (formerly <i>SEPN1</i> ) 606210	Selenoprotein N	Moghadaszadeh et al (1998, 2001) Ferreiro et al (2002, 2004)	
Congenital muscular dystrophy due to ITGA7 deficiency	2.14	AR	613204	12q13.2	<b>ITGA7</b> 600536	Integrin alpha7	Hayashi et al (1998)	
Congenital muscular dystrophy related to <i>DNM2</i>	2.15	AD		19p13.2	<b>DNM2</b> 602378	Dynamin 2	Susman et al (2010)	allelic to CNM1 (group 3), Distal myopathy related to <i>DNM2</i> (group 4), LCCS5 (group 12) and CMTDIB (group 14)
Congenital muscular dystrophy related to <i>TCAP</i>	2.16	AR		17q12	<b>TCAP</b> 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	<b>LMNA</b> 150330	Lamin A/C	Quijano-Roy et al (2008)	Allelic to EDM2, EDM3 (group 1), CMD1A (group 10A), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermatopathy #275210, MADA #248370]
Rigid spine syndrome related to <i>HMGCS1</i>	2.18	AR		5p12	<b>HMGCS1</b> 142940	3-hydroxy-3-methylglutaryl-coa synthase 1	Dofash et al (2024)	
<b>CMD-Dystroglycanopathies</b>								
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A1 formerly Walker-Warburg syndrome (WWS)	2.19	AR	MDDGA1 236670	9q34.13	<b>POMT1</b> 607423	Protein-O-mannosyl transferase 1	Beltran-Valero De Bernabe et al (2002) Mercuri et al (2009)	allelic to MDDGC1 (group 1), and MDDGB1 (group 2)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A2 formerly Walker-Warburg syndrome (WWS)	2.20	AR	MDDGA2 613150	14q24.3	<b>POMT2</b> 607439	Protein O-mannosyl transferase 2	van Reeuwijk et al (2005) Mercuri et al (2009)	Allelic to MDDGC2 (group 1) and MDDGA2, MDDGB2 (group 2)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A3 formerly Walker-Warburg syndrome (WWS)	2.21	AR	MDDGA3 253280	1p34.1	<b>POMGNT1</b> 606822	Protein O-mannose beta1,2-N-acetylglucosaminyl transferase	Yoshida et al (2001) Taniguchi et al (2003)	Allelic to MDDGC3 (group 1) and MDDGB3 (group 2)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A4 formerly Fukuyama congenital muscular dystrophy (FCMD)	2.22	AR	MDDGA4 253800	9q31.2	<b>FKTN</b> 607440	Fukutin	Toda et al. (1993) Kobayashi et al (1998)	Allelic to MDDGC4 (group 1), MDDGB4 (group 2) and CMD1X (group 10A)

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Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A5 formerly Walker-Warburg syndrome (WWS)	2.23	AR	MDDGA5 613153	19q13.32	<b>FKRP</b> 606596	Fukutin related protein	Cormand et al (2001) Beltran-Valero De Bernabe et al (2004)	Allelic to MDDGC5 (group 1) and MDDGB5 (group 2)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A6	2.24	AR	MDDGA6 613154	22q12.3	<b>LARGE</b> 603590	Acetylglucosaminyl-transferase-like protein (like-glycosyl transferase)	Longman et al (2003) Mercuri et al (2009)	Allelic to MDDGB6 (group 2)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A7 formerly Walker-Warburg syndrome (WWS)	2.25	AR	MDDGA7 614643	7p21.2	<b>CRPPA</b> 614631	Isoprenoid synthase domain containing	Roscioli et al (2012) Willer et al (2012)	Allelic to MDDGC7 (group 1)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A8 formerly Walker-Warburg syndrome (WWS)	2.26	AR	MDDGA8 614830	3p22.1	<b>POMGNT2</b> 614828	Protein O-mannose beta-1,4-N-acetylglucosaminyl transferase 2	Manzini et al (2012)	allelic to MDDGC8 (group 1)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A9	2.27	AR	MDDGA9 616538	3p21.31	<b>DAG1</b> 128239	Dystrophin-Associated Glycoprotein 1	Geis et al (2013)	allelic to MDDGC9 (group 1)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A10	2.28	AR	MDDGA10 615041	12q14.2	<b>RXYLT1</b> 605862	Ribitol xylosyltransferase 1 (transmembrane protein 5)	Vuillaumier-Barrot et al (2013)	
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A11	2.29	AR	MDDGA11 615181	1q42.3	<b>B3GALNT2</b> 610194	Beta-1,3-N-acetylgalactosaminyl-transferase 2	Stevens et al (2013)	
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A12	2.30	AR	MDDGA12 615249	8p11.21	<b>POMK</b> 615247	Protein-O-mannose kinase	Jae et al (2013)	Allelic to MDDGC12 (group 1)
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A13 formerly Walker-Warburg syndrome (WWS)	2.31	AR	MDDGA13 615287	11q13.2	<b>B4GAT1</b> 605517	Beta-1,4-glucuronyl transferase 1 (Beta-1,3-N-acetylglucosaminyl-transferase 1)	Buysse et al (2013) Shaheen et al (2013)	
Muscular dystrophy-dystroglycanopathy (Congenital with brain and eyes anomalies), type A14 formerly Walker-Warburg syndrome (WWS) and Muscle Eye Brain	2.32	AR	MDDGA14 615350	3p21.31	<b>GMPPB</b> 615320	GDP-mannose pyrophosphorylase B	de Carss et al (2013)	Allelic to MDDGC14 (group 1), MDDGB14 (group 2), congenital myasthenic syndrome (group 11)
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B1	2.33	AR	MDDGB1 613155	9q34.13	<b>POMT1</b> 607423	Protein-O-mannosyl transferase 1	van Reeuwijk et al (2006) Mercuri et al (2009)	Allelic to MDDGC1 (group 1) and MDDGA1 (group 2)
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B2 formerly Walker-Warburg syndrome (WWS)	2.34	AR	MDDGB2 613156	14q24.3	<b>POMT2</b> 607439	Protein O-mannosyl transferase 2	Yanagisawa et al (2007)	Allelic to MDDGC2 (group 1) and MDDGA2 and MDDGB2 (group 2)
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B3 formerly Walker-Warburg syndrome (WWS)	2.35	AR	MDDGB3 613151	1p34.1	<b>POMGNT1</b> 606822	Protein O-mannose beta1,2-N-acetylglucosaminyl transferase	Clement et al (2008) Mercuri et al (2009)	Allelic to MDDGC3 (group 1) and MDDGA3 (group 2)

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Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B4 formerly Walker-Warburg syndrome (WWS)	2.36	AR	MDDGB4 613152	9q31.2	<i>FKTN</i> 607440	Fukutin	Beltran-Valero de Bernabe (2003) Mercuri et al (2009)	Allelic to MDDGC4 (group 1), MDDGA4 (group 2) and CMD1X (group 10A)
Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B5	2.37	AR	MDDGB5 606612	19q13.32	<i>FKRP</i> 606596	Fukutin related protein	Brockington et al (2001b) Topaloglu et al (2003) Mercuri et al (2009)	Allelic to MDDGC5 (group 1) and MDDGA5 (group 2)
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B6	2.38	AR	MDDGB6 608840	22q12.3	<i>LARGE</i> 603590	Acetylglucosaminyl-transferase-like protein (like-glycosyl transferase)	Longman et al (2003) Mercuri et al (2009)	Allelic to MDDGA6 (group 2)
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B14	2.39	AR	MDDGB14 615351	3p21.31	<i>GMPPB</i> 615320	GDP-mannose pyrophosphorylase B	Carss et al (2013)	Allelic to MDDGC14 (group 1), MDDGA14 (group 2) and congenital myasthenic syndrome (group 11)
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B15	2.40	AR	MDDGB15 618992	1q22	<i>DPM3</i> 605951		Fu et al (2019)	Allelic to MDDGC15 (group 1)
Muscle-eye-brain disease (MEB)	2.41	AD	MEB	11q13.2q14.1	<i>11q13.2q14.1 duplication TRAPPC11</i> 614138		Villar-Quiles et al (2020)	
Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by <i>TRAPPC11</i> mutations	2.42	AR		4q35.1		Trafficking protein particle complex 11	Liang et al (2015) Larson et al (2018)	allelic to LGMDR18 (group 1)
Congenital muscular dystrophy, with or without seizures	2.43	AR	MYOS 620166	17q21.32	<i>GOSR2</i> 604027	Golgi SNAP receptor complex member 2	Tsai et al (2013) Larson et al (2018)	
<b>Other congenital muscular dystrophies</b>								
Congenital muscle dystrophy with mitochondrial structural abnormalities (megaconial type)	2.44	AR	MDCMC 602541	22q13.33	<i>CHKB</i> 612395	Choline kinase beta	Mitsuhashi et al (2011)	
Congenital muscular dystrophy	2.45	AR	MDC1B 604801	1q42	?		Brockington et al (2000)	
Congenital muscular dystrophy with rigid spine related to <i>ACTA1</i>	2.46	AR	Possibly identical to MDC1B 604801	1q42.13	<i>ACTA1</i> 102610	Alpha actin, skeletal muscle	O'Grady et al. (2014)	Allelic to CMYO2A, CMYO2B, CMYO2C, CFTD and cap myopathy (group 3)
Developmental delay with hypotonia, myopathy and brain abnormalities	2.47	AR	DEDHMB 620240	9q34.11	<i>GOLGA2</i> 602580	Golgin A2	Shamseldin et al (2016)	
Muscular dystrophy, congenital Davignon-Chauveau type	2.48	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	2.49	AR	MDCCAIID 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>	2.50	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)

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Congenital muscular dystrophy related to <i>RYR1</i>	2.51	AR		19q13.2	<b><i>RYR1</i></b> 180901	Ryanodine receptor	Helbling et al (2019)	Allelic to CMYO1A, CMYO1B, CCD, CFTD, CNMDU1, Centronuclear myopathy related to RYR1, DuCD related to RYR1 (group 3), Late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia deformation sequence related to RYR1 (group 16)
Congenital muscular dystrophy related to <i>TRAPPC2L</i>	2.52	AR		16q24.3	<b><i>TRAPPC2L</i></b> 610970	Trafficking protein particle complex 2-like	Abaji et al (2023)	
<b>GROUP 3. CONGENITAL MYOPATHIES</b>								
<b><i>Nemaline myopathies</i></b>								
Congenital myopathy 2A, typical Formerly Nemalin myopathy 3	3.1	AD	CMYO2A 161800	1q42.13	<b><i>ACTA1</i></b> 102610	Actin, alpha 1, skeletal muscle	Agrawal et al (2004)	Allelic to CMD with rigid spine (group 2), CMYO2B, CMYO2C, CFTD and cap myopathy (group 3)
Congenital myopathy 2B, severe infantile	3.2	AR	CMYO2B 161800	1q42.13	<b><i>ACTA1</i></b> 102610	Actin, alpha 1, skeletal muscle	Nowak et al (1999) Sparrow et al (2003)	Allelic to CMD with rigid spine (group 2), CMYO2A, CMYO2C, CFTD and cap myopathy (group 3)
Congenital myopathy 4A, dominant Formerly Nemalin myopathy 1	3.3	AD	CMYO4A 255310	1q21.3	<b><i>TPM3</i></b> 191030	Tropomyosin 3	Laing et al. (1992) Laing et al. (1995b) Tan et al (1999) Wattanasirichaigoon et al (2002)	Allelic to CMYO4B (group 3)
Congenital myopathy 4B, recessive	3.4	AR	CMYO4B 609284	1q21.3	<b><i>TPM3</i></b> 191030	Tropomyosin 3	Tan et al (1999) Wattanasirichaigoon et al (2002)	Allelic to CMYO4A (group 3)
Nemalin myopathy 2	3.5	AR	NEM2 256030	2q23.3	<b><i>NEB</i></b> 161650	Nebulin	Wallgren-Petersson et al (1995, 2002) Pelin et al (1999) Lehtokari et al (2006)	allelic to Distal myopathy with nebulin defect (group 4)
Nemalin myopathy 4	3.6	AD	CMYO23 609285	9p13.3	<b><i>TPM2</i></b> 190990	Tropomyosin 2 (beta)	Donner et al (2002) Monnier et al (2009)	Allelic to CMYO23 (group 3), DA2B4 and DA1A (group 16)
Nemalin myopathy 5	3.7	AR	NEM5A 605355	19q13.42	<b><i>TNNT1</i></b> 191041	Troponin T type 1 (skeletal, slow)	Johnston et al (2000)	
Nemalin myopathy 6	3.8	AD	NEM6 609273	15q22.31	<b><i>KBTBD13</i></b> 613727	Kelch repeat and BTB/POZ domain containing protein 13	Gommans et al (2003) Samburghin et al (2010)	allelic to LGMD related to <i>KBTBD13</i> (group 1)
Nemalin myopathy 7	3.9	AR	NEM7 610687	14q13.1	<b><i>CFL2</i></b> 601443	Cofilin 2 (muscle)	Agrawal et al (2007)	
Nemalin myopathy 8	3.10	AR	NEM8 615348	3p22.1	<b><i>KLHL40</i></b> 615340	Kelch-like family member 40	Ravenscroft et al (2013)	
Nemalin myopathy 9	3.11	AR	NEM9 615731	2q31.1	<b><i>KLHL41</i></b> 607701	Kelch-like family member 41	Gupta et al (2013)	
Nemalin myopathy 10	3.12	AR	NEM10 616165	3p14.1	<b><i>LMOD3</i></b> 616112	Leiomodin 3	Yuen et al (2014)	
Nemalin myopathy 11	3.13	AR	CMYO24 617336	10q21.3	<b><i>MYPN</i></b> 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	3.14	AR	KFS4 616549	22q12.1	<b><i>MYO18B</i></b> 607295	Myosin XVIIIIB	Malfatti et al (2015)	
Congenital myopathy 20	3.15	AR	CMYO20 620310	15q13-q14	<b><i>RYR3</i></b> 180903	Ryanodine receptor 3	Nilipour et al (2018)	
Nemalin myopathy with distal arthrogyposis	3.16	AR		11p15.5	<b><i>TNNT3</i></b> 600692	Troponin T3, fast skeletal	Sandaradura et al (2018)	Allelic to DA2B2 (group 16)
<b><i>Other congenital myopathies</i></b>								
Myotubular myopathy, X-linked	3.17	XLR	CNMX 310400	Xq28	<b><i>MTM1</i></b> 300415	Myotubularin 1	Thomas et al (1987) Laporte (1996, 1997, 2000)	
Centronuclear myopathy 1	3.18	AD	CNM1 160150	19p13.2	<b><i>DNM2</i></b> 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	3.19	AR, AD	CNM2 255200	2q14.3	<b><i>BINI</i></b> 601248	Bridging integrator 1 (Amphiphysin)	Nicot et al (2007) Böhm et al (2014)	
Centronuclear myopathy 4	3.20	AD	CNM4 614807	16p13.3	<b><i>CCDC78</i></b> 614666	Coiled-coil domain-containing protein 78	Majczenko et al (2012)	
Centronuclear myopathy 5	3.21	AR	CNM5 615959	2q35	<b><i>SPEG</i></b> 615950	SPEG complex locus	Agrawal 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)
Myopathy centronuclear with fiber-type disproportion	3.22	AR	CNM6 617760	3p23-21	<b>MAP3K20</b> 609479	Mitogen-activated protein 3 kinase 20	Tetreault et al (2006) Vasli et al (2017)	
Centronuclear myopathy related to <i>RYR1</i>	3.23	AR		19q13.2	<b>RYR1</b> 180901	Ryanodine receptor	Wilmshurst et al (2010)	Allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYO1A</i> , <i>CMYO1B</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16)
Centronuclear myopathy (CNM) related to <i>TTN</i>	3.24	AR		2q31.2	<b>TTN</b> 188840	Titin	Ceyhan-Birsoy et al (2013)	Allelic to <i>LGMDR10</i> (group 1), <i>MmD</i> related to <i>TTN</i> and <i>CMYO5</i> (group 3), <i>TMD</i> (group 4), <i>MFM9</i> (group 5), <i>CMH9</i> (group 10), <i>CMD1G</i> (group 10), <i>LCCS</i> related to <i>TTN</i> (group 12)
Congenital myopathy 1A, dominant, with susceptibility to malignant hyperthermia	3.25	AD	<i>CMYO1A</i> 117000	19q13.2	<b>RYR1</b> 180901	Ryanodine receptor	Kausch et al (1991) Zhang et al (1993) Quane et al. (1993) Robinson et al (2002)	Allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYO1B</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16)
Central core disease, recessive (transient multiminicore myopathy)	3.26	AR	<i>CMYO1A</i> 117000	19q13.2	<b>RYR1</b> 180901	Ryanodine receptor	Ferreiro et al (2002a) Jungbluth et al (2002)	Allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYO1A</i> , <i>CMYO1B</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16)
Congenital myopathy 1B, recessive Minicore myopathy with external ophthalmoplegia	3.27	AR	<i>CMYO1B</i> 255320	19q13.2	<b>RYR1</b> 180901	Ryanodine receptor	Monnier et al (2003) Jungbluth et al (2005)	Allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYO1A</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , <i>DuCD</i> related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8), Fetal akinesia deformation sequence related to <i>RYR1</i> (group 16)
Multiminicore disease ( <i>MmD</i> ) related to <i>TTN</i>	3.28	AR		2q31.2	<b>TTN</b> 188840	Titin	Ge et al (2019)	Allelic to <i>LGMDR10</i> (group 1), <i>CNM</i> related to <i>TTN</i> and <i>CMYO5</i> (group 3), <i>TMD</i> (group 4), <i>MFM9</i> (group 5), <i>CMH9</i> (group 10A), <i>CMD1G</i> (group 10A), <i>LCCS</i> related to <i>TTN</i> (group 12)
Congenital myopathy 10A, severe variant	3.29	AR	<i>CMYO10A</i> 614399	5q23.2	<b>MEGF10</b> 612453	Multiple EGF-like-domains 10	Logan et al (2011)	Allelic to <i>CMYO10B</i> (group 3)
Congenital myopathy 10B, mild variant	3.30	AR	<i>CMYO10B</i> 620249	5q23.2	<b>MEGF10</b> 612453	Multiple EGF-like-domains 10	Boyden et al (2012)	Allelic to <i>CMYO10A</i> (group 3)
Congenital myopathy 7A, myosin storage, autosomal dominant	3.31	AD	<i>CMYO7A</i> 608358	14q11.2	<b>MYH7</b> 160760	Myosin, heavy chain 7, cardiac muscle, beta	Tajsharghi et al (2003) Bohlega et al (2004) Laing et al (2005)	Allelic to <i>CFTD</i> , <i>CMYO7B</i> , <i>MSM</i> with eccentric core disease (group 3), <i>MPD1</i> (group 4), <i>CMH1</i> , <i>CMD1S</i> and <i>LVNC5</i> (group 10A)
Congenital myopathy 7B, myosin storage, autosomal recessive	3.32	AR	<i>CMYO7B</i> 255160	14q11.2	<b>MYH7</b> 160780	Myosin, heavy chain 7, cardiac muscle, beta	Onengut et al (2004) Tajsharghi et al (2007) Yuceyar et al (2015)	Allelic to <i>CFTD</i> , <i>CMYO7A</i> , <i>MSM</i> with eccentric core disease and <i>SPMM</i> related to <i>MYH7</i> (group 3), <i>MPD1</i> (group 4), <i>CMH1</i> , <i>CMD1S</i> and <i>LVNC5</i> (group 10A)
Myopathy, myosin storage, autosomal dominant (eccentric core disease)	3.33	AD		14q11.2	<b>MYH7</b> 160760	Myosin, heavy chain 7, cardiac muscle, beta	Romero et al (2014)	Allelic to <i>CFTD</i> , <i>CMYO7A</i> , <i>CMYO7B</i> (group 3), <i>MPD1</i> (group 4), <i>CMH1</i> , <i>CMD1S</i> and <i>LVNC5</i> (group 10A)

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Congenital myopathy 6 with ophtalmoplegia	3.34	AD, AR	CMYO6 605637	17p13.1	<b>MYH2</b> 160740	Myosin, heavy chain 2, skeletal muscle, adult	Martinsson et al (1999, 2000) Tajsharghi et al (2010)	
Isolated inclusion body myopathy	3.35	AD	IBMPFD3 615424	12q13.13	<b>HNRNPA1</b> 164017	Heterogeneous nuclear ribonucleoprotein A1	Izumi et al (2015)	allelic to MPD3 (group 4) and ALS20 (group 12)
Congenital myopathy 23	3.36	AD	CMYO23 609285	9p13.3	<b>TPM2</b> 190990	Tropomyosin 2, beta	Tajsharghi et al (2007) Lehtokari et al (2007)	Allelic to NEM4 (group 3), DA1A and DA2B4 (group 16)
Congenital myopathy 2C, severe infantile, dominant	3.37	AD	CMYO2C 620278	1q42.13	<b>ACTA1</b> 102610	Actin, alpha 1, skeletal muscle	Goebel et al (1997) Nowak et al (1999)	Allelic to CMD with rigid spine (group 2), CMYO2A, CMYO2B, CMYO2C, CFTD and cap myopathy (group 3)
Cap myopathy	3.38	AD		1q42.13	<b>ACTA1</b> 102610	Actin, alpha 1, skeletal muscle	Hung et al (2010)	Allelic to CMD with rigid spine (group 2), CMYO2A, CMYO2B, CMYO2C and CFTD (group 3)
Congenital myopathy 5 with cardiomyopathy Formerly Salih myopathy (Congenital myopathy with fatal cardiomyopathy)	3.39	AR	CMYO5 611705	2q31.2	<b>TTN</b> 188840	Titin	Carmignac et al (2007)	Allelic to LGMDR10 (group 1), CNM related to TTN and MmD related to TTN (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10), CMD1G (group 10), LCCS related to TTN (group 12)
Congenital skeletal myopathy and fatal cardiomyopathy	3.40	AR		11p11.2	<b>MYBPC3</b> 600958	Cardiac myosin binding protein-C	Tajsharghi et al (2010)	allelic to CMH4, CMD1MM and LVNC10 (group 10A)
Congenital myopathy 12 Formerly Congenital myopathy Compton-North Sarcotubular myopathy	3.41	AR	CMYO12 612540	12q12	<b>CNTN1</b> 600016	Contactin-1	Compton et al (2008)	
	3.42	AR		9q33.1	<b>TRIM32</b> 602290	Tripartite motif containing 32	Schoser et al (2005)	allelic to LGMDR8 (group 1)
Congenital Myopathy 11	3.43	AR	CMYO11 619967	10p12.33	<b>HACD1</b> (PTPLA) 610467	3-Hydroxyacyl-CoA dehydratase (Protein tyrosine phosphatase-like)	Muhammad et al (2013)	allelic to ARVD6 (group 10B)
Congenital myopathy with ophthalmoplegia related to CACNA1S	3.44	AR		1q32.1	<b>CACNA1S</b> 114208	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Hunter et al. (2015)	Allelic to LGMD related to CACNA1S (group 1), CMYO18 (group 3), HOKPP1 (group 7) and MHS5 (group 8)
Neurodevelopmental disorder with hypotonia, neuropathy and deafness	3.45	AR	NEDHND 617519	19q13.2	<b>SPTBN4</b> 606214	Spectrin, beta, nonerythrocytic, 4	Knierim et al (2017)	
Myopathy, congenital, with excess of muscle spindles	3.46	AD	CSTLO 218040	11p15.5	<b>HRAS</b> 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)	3.47	AR	CFZS 254940	9q34.2	<b>MYMK</b> 615345	Myomaker	Di Gioia et al (2017)	
Congenital myopathy 8	3.48	AD	CMYO8 618654	1q43	<b>ACTN2</b> 102573	Actinin, alpha 2	Lornage et al (2019)	Allelic to MPD6 (group 4), CMH23 and CMD1AA (group 10A)
Congenital myopathy 9A	3.49	AR	CMYO9A 618822	3q26.33	<b>FXR1</b> 600819	FMR1 autosomal homolog	Estan et al (2019)	Allelic to CMYO9B (group 3)
Congenital myopathy 9B, proximal with minicore	3.50	AR	CMYO9B 618823	3q26.33	<b>FXR1</b> 600819	FMR1 autosomal homolog	Estan et al (2019)	Allelic to CMYO9A (group 3)
Congenital myopathy 13	3.51	AR	CMYO13 255995	12q13.3	<b>STAC3</b> 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	3.52	AR	CMYO13 255995	12q13.3	<b>STAC3</b> 615521	SH3 and cysteine-rich domains 3	Zaharieva et al (2018)	Allelic to CMYO13 (group 3)
Congenital myopathy 14	3.53	AR	CMYO14 618414	2q34	<b>MYL1</b> 160780	Myosin, light polypeptide 1, alkali, skeletal fast	Ravenscroft et al (2018)	
Congenital myopathy 15	3.54	AD	CMYO15 620161	20q13.12	<b>TNNC2</b> 191039	Troponin C Fast	van de Loch (2021)	
Congenital myopathy 16	3.55	AD	CMYO16 618524	12q23.2	<b>MYBPCI</b> 160794	Myosin-binding protein C, slow type	Shashi et al (2019)	Allelic to LCCS4 (group 12), DA1B (group 16)
Congenital myopathy 17	3.56	AR	CMYO17 618975	11p15.1	<b>MYOD1</b> 159970	Myogenic Differentiation Antigen 1	Watson et al (2016)	

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Congenital myopathy 18 due to dihydropyridine receptor defect	3.57	AD, AR	CMYO18 620246	1q32.1	<i>CACNA1S</i> 114208	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Schartner et al (2019)	Allelic to LGMD related to CACNA1S (group 1), Congenital myopathy with ophthalmoplegia (group 3), HOKPP1 (group 7) and MHS5 (group 8)
Congenital myopathy 19	3.58	AR	CMYO19 618578	1p36.13	<i>PAX7</i> 167410	Paired Box gene 7	Feichtinger et al (2019)	
Congenital Myopathy 22A, classic	3.59	AR	CMYO22A 620351	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 CMYO22B (group 3), HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16)
Congenital Myopathy 22B, severe fetal	3.60	AR	CMYO22B 620369	17q23.3	<i>SCN4A</i> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Zaharieva et al (2016)	Allelic to CMYO22A (group 3), HYPP, HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16)
Congenital Myopathy related to <i>PYROXD1</i>	3.61	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>	3.62	AD		7q32.1	<i>TNPO3</i> 610032	Transportin 3	Vihola et al (2019) Angelini et al (2019)	allelic to LGMDD2 (group 1)
Congenital amyotrophy	3.63	AR		16p13.3	<i>CACNA1H</i> 607904	Calcium channel, voltage-dependent, T type, alpha-1H subunit	Carter 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 Congenital muscular dystrophy related to RYR1 (group 2), CMYO1A, CMYO1B, CCD, CFTD, CNMDU1, Centronuclear myopathy related to RYR1 (group 3), Late onset axial myopathy (group 5), MHS1 (group 8), Fetal akinesia deformation sequence related to RYR1 (group 16)
Lysosomal storage myopathy	3.65	AR	ML4 252650	19p13.2	<i>MCOLN1</i> 605248	Mucopolin 1	Zambon et al. (2021)	
Congenital myopathy 25	3.66	AR	CMYO25 620964	8q21.11	<i>JPH1</i> 605266	Junctophilin 1	Johari et al (2024)	
Congenital myopathy with Skeletal Deformities, Intellectual Disability, and Ovarian Dysfunction	3.67	AR		16p13.3	<i>SOX8</i> 605923	SRY-Box 8	Warman-Chardon et al (2023)	

**GROUP 4. DISTAL MYOPATHIES**

Miyoshi muscular dystrophy 1	4.1	AR	MMD1 254130	2p13.2	<i>DYSF</i> 603009	Dysferlin	Bejaoui et al (1995) Bashir et al (1998) Liu et al (1998)	allelic to LGMDR2 (group 1)
Miyoshi muscular dystrophy 2	4.2	AR	MMD2 613318	8q22.3			Linssen et al (1998)	
Miyoshi muscular dystrophy 3	4.3	AR	MMD3 613319	11p14.3	<i>ANO5</i> 608662	Anoctamin 5	Bolduc et al. (2010)	allelic to LGMDR12 (group 1) and Muscular dystrophy with gnathodiaphyseal dysplasia (group 1)
Tibial muscular dystrophy (Udd)	4.4	AD	TMD 600334	2q31.2	<i>TTN</i> 188840	Titin	Haravuori et al (1998, 2001) Hackman et al (2002)	Allelic to LGMDR10 (group 1), CNM related to TTN, MmD related to TTN and CMYO5 (group 3), MFM9 (group 5), CMH9 (group 10), CMD1G (group 10), LCCS related to TTN (group 12)
Nonaka Myopathy	4.5	AR	NM 605820	9p13.3	<i>GNE</i> 603824	Glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase	Mitrani-Rosenbaum et al (1996) Ikeuchi et al (1997) Eisenberg et al (2001)	

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Myopathy, Distal, 1 (Laing)	4.6	AD	MPD1 160500	14q11.2	<b>MYH7</b> 160760	Myosin, heavy chain 7, cardiac muscle, beta	Laing et al. (1995) Mastaglia et al (2000) Meredith et al (2004)	Allelic to CFTD, CMYO7A, CMYO7B, MSM with eccentric core disease (group 3), CMH1, CMD1S and LVNC5 (group 10A)
Myopathy distal 3	4.7	AD	MPD3 610099	12q13-q22	<b>HNRNPA1</b> 164017	Heterogeneous nuclear ribonucleoprotein A1	Haravuori et al (2004) Hackman et al (2021)	Allelic to IBMPFD3 (group 3) and ALS20 (group 12)
Myopathy, distal, 4	4.8	AD	MPD4 614065	7q32.1	<b>FLNC</b> 102565	Filamin C (gamma)	Duff et al (2011)	allelic to MFM5 (group 5), CMH26 and RCM5 (group 10A)
Myopathy, distal, 5	4.9	AR	MPD5 617030	14q32.33	<b>ADSS1</b> 612498	Adenylosuccinate synthase 1	Park et al (2016)	
Myopathy, distal 6, Adult-onset	4.10	AD	MPD6 618655	1q43	<b>ACTN2</b> 102573	Actinin alpha-2	Savarese et al (2019)	Allelic to CMYO8 (group 3), CMH23 and CMD1AA (group 10A)
Myopathy, distal, 7 adult onset, X-linked	4.11	XLD	MPD7 301075	Xp22.12	<b>SMPX</b> 300226	Small Muscle Protein, X-linked	Johari et al (2021)	
Myopathy distal, Tateyama type	4.12	AD	MPDT 614321	3p25.3	<b>CAV3</b> 601253	Caveolin-3	Tateyama et al (2002) Fulizio et al (2005)	allelic to formerly LGMD1C (group 1), Creatin phosphokinase elevated serum (group 5), RMD2 (group 6), CMH1 (group 10A) and LQT9 (group 10B)
Vocal cord and pharyngeal distal myopathy (VCPDM) reclassified as ALS21	4.13	AD	ALS21 606070	5q31.2	<b>MATR3</b> 164015	Matrin 3	Feit et al (1998) Senderek et al (2009)	Allelic to ALS21 (group 12)
Welander distal myopathy	4.14	AD	WDM 604454	2p13.3	<b>TIA1</b> 603518	Cytotoxic granule-associated RNA binding protein	Ahlberg et al (1999) Hackman et al (2013) Klar et el (2013) Lee et al (2018)	allelic to Welander-like syndrome (group 4) and ALS26 (group 12)
Welander-like distal myopathy	4.15	Digenic		5q35.3 2p13.3	<b>SQSTM1</b> 601530 + <b>TIA1</b> 603518	1 + Cytotoxic granule-associated RNA binding protein		<b>SQSTM1</b> Allelic to DMRV (group 4) and FTDALS3 (group 12) <b>TIA1</b> Allelic to WDM (group 4) and ALS26 (group 12)
Distal myopathy with myotilin defect	4.17	AD		5q31.2	<b>MYOT</b> 604103	Myotilin	Penisson-Besnier et al (1998, 2006)	Allelic to MFM3 (group 5), spheroid body myopathy (group 5)
Distal myopathy with nebulin defect	4.18	AR		2q23.3	<b>NEB</b> 161650	Nebulin	Wallgren-Petersson et al (2007)	allelic to NEM2 (group 3) and AD Distal myopathy with nebulin defect (group 4)
Distal myopathy with nebulin defect	4.19	AD		2q23.3	<b>NEB</b> 161650	Nebulin	Kiiski et al (2019)	allelic to NEM2 (group 3) and AR Distal myopathy with nebulin defect (group 4)
Late onset distal myopathy (Markesbery-Griggs)	4.20	AD		10q23.2	<b>LDB3</b> 605906	LIM domain binding-3	Griggs et al (2007)	allelic to MFM4 (group 5), CMD1C, CMH24 and LVNC3 (group 10A)
Distal myopathy related to <i>DNM2</i>	4.21	AD		19p13.2	<b>DNM2</b> 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	4.22	AD		9p21.3	<b>KLHL9</b> 611201	Kelch-like 9	Cirak et al (2010)	
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 1	4.23	AD	IBMPFD1 167320	9p13.3	<b>VCP</b> 601023	Valosin-containing protein	Palmio et al (2011)	allelic to Scapuloperoneal myopathy (group 1), IBMPFD1 (group 5), ALS14 (group 12) and CMT2Y (group 14)
Myopathy, distal, with rimmed vacuoles	4.24	AD	DMRV 617158	5q35.3	<b>SQSTM1</b> 601530	Sequestosome 1	Bucelli et al (2015)	allelic to Welander-like syndrome (group 4) and FTDALS3 (group 12)
Myopathy, distal, with rimmed vacuoles	4.25	AD		7q36.3	<b>DNAJB6</b> 611332	Hsp40 homologue, subfamily B, number 6	Ruggieri et al (2015)	allelic to LGMDD1 (group 1) and distal myopathy (group 4)
Distal myopathy	4.26	AD		7q36.3	<b>DNAJB6</b> 611332	Hsp40 homologue, subfamily B, number 6	Palmio et al (2019)	Allelic to LGMDD1 (group 1), Rimmed vacuole distal myopathy (group 4)
Distal Myopathy	4.27	AD		19p13.3	<b>PLIN4</b> 613247	Perilipin 4	Ruggieri et al (2020)	
Rimmed vacuole myopathy	4.28	AD		12q24.23	<b>HSPB8</b> 608014	Heat-shock 22kD protein 8	Al-Tahan et al (2019)	Allelic to distal myopathy and motor neuropathy (group 4) HMND2 (group 12) and CMT2L (group 14)

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Distal Myopathy and motor neuropathy	4.29	AD		12q24.23	<b>HSPB8</b> 608014	Heat-shock 22kD protein 8	Ghaoui et al (2016)	Allelic to Rimmed vacuole myopathy (group 4), HMN2A (group 12) and CMT2L (group 14)
Distal myopathy, late-onset	4.30	AD		1p36.22	<b>TARDBP</b> 605078	Tar DNA-binding protein	Zibold et al (2024)	Allelic to ALS10 (group 12)

**GROUP 5. OTHER MYOPATHIES***Myofibrillar myopathies*

Myofibrillar myopathy 1	5.1	AD, AR	MFM1 601419	2q35	<b>DES</b> 125660	Desmin	Goldfarb et al (1998) Munoz-Marmol et al (1998)	Allelic to formerly LGM2R (group 1), MFM1 with ARCV7 (group 5), CMD11 (group 10A), ARVC7 (group 10B), myopathy, cardiomyopathy and CMS (group 11)
Myofibrillar myopathy 2	5.2	AD	MFM2 608810	11q23.1	<b>CRYAB</b> 123590	Alpha-B crystallin	Vicart et al (1998) Selcen et al (2003)	Allelic to CMD11I (group 10A) and CMT2 related to <b>CRYAB</b> (group 14)
Myofibrillar myopathy 3	5.3	AD	MFM3 609200	5q31.2	<b>MYOT</b> 604103	Myotilin	Selcen and Engel (2004)	Allelic to distal myotilinopathy (group 4), spheroid body myopathy (group 5)
Spheroid body myopathy	5.4	AD	182920	5q31.2	<b>MYOT</b> 604103	Myotilin	Foroud et al (2005)	allelic to distal myotilinopathy (group 4), MFM3 (group 5)
Myofibrillar myopathy 4	5.5	AD	MFM4 609452	10q23.2	<b>LDB3</b> 605906	LIM domain binding-3	Selcen and Engel (2005)	allelic to Late onset distal myopathy (group 4), CMD1C, CMH24 and LVNC3 (group 10A)
Myofibrillar myopathy 5	5.6	AD, AR	MFM5 609524	7q32.1	<b>FLNC</b> 102565	Filamin C (gamma)	Vorgerd et al (2005) Kölbl et al (2020) Schuld et al (2020)	allelic to MPD4 (group 4), CMH26 and RCM5 (group 10A)
Myofibrillar myopathy 6	5.7	AD	MFM6 612954	10q26.11	<b>BAG3</b> 603883	BCL2-associated athanogene 3	Selcen et al (2009)	allelic to CMD11H (group 10A) and CMT related to BAG3 (group 14)
Myofibrillar myopathy 7	5.8	AR	MFM7 617114	3q22.2	<b>KY</b> 605739	Kyphoscoliosis peptidase	Hedberg-Oldfors et al (2016) Straussberg et al (2016)	
Myofibrillar myopathy 8	5.9	AR	MFM8 617258	12p12.1	<b>PYROXDI</b> 617220	Pyridine nucleotide-disulphide oxidoreductase domain 1	O'Grady et al (2016)	allelic to LGMD related to <b>PYROXDI</b> (group 1) and Congenital Myopathy related to <b>PYROXDI</b> (group 3)
Myofibrillar myopathy 9 with early respiratory failure	5.10	AD	MFM9 603689	2q31.2	<b>TTN</b> 188840	Titin	Nicolao et al (1999) Lange et al (2005)	Allelic to LGMDR10 (group 1), CNM related to TTN, MmD related to TTN and CMYO5 (group 3), TMD (group 4), CMH9 (group 10A), CMD1G (group 10A), LCCS related to TTN (group 12)
Myofibrillar myopathy 10	5.11	AR	MFM10 619040	10p11.23	<b>SVIL</b> 604126	Supervillin	Hedberg-Oldfors et al (2020)	
Myofibrillar myopathy 11	5.12	AR	MFM11 619178	17q12	<b>UNC45B</b> 611220	UNC45 Myosin Chaperone B	Dafsari et al (2019)	
Myopathy, myofibrillar 12, infantile onset, with cardiomyopathy	3.20	AR	MFM12 619424	12q24.11	<b>MYL2</b> 160781	Myosin regulatory light chain 2	Weterman et al (2013)	Allelic to CMH10 (group 10A)
Cardiac and skeletal aggregate myopathy	5.14	Digenic		1p36.11 2p.23.3	<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	5.15	AD	MFM1 601419	10q22 > 2q35	<b>DES</b> 125660	Desmin	Melberg et al (1999) Kuhl et al (2008) Hedberg et al (2012)	Allelic to MFM1 (group 5), CMD11 (group 10A), ARVC7 (group 10B) and myopathy, cardiomyopathy and CMS (group 11)
Congenital myopathy 21 with early respiratory failure	5.16	AR	CMYO21 620326	1p31.1	<b>DNAJB4</b> 611327	DNAJ/HSP40 homolog, subfamily B, member 4	Wheil et al (2022)	
Neuromuscular disorder, congenital, with dysmorphic facies	5.17	AR	NMDF 620775	6q14.1	<b>FILIP1</b> 607307	Filamin A-interacting protein 1	Schnabel et al (2023) Roos et al (2023)	

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<b>Miscellaneous</b>								
Danon disease	5.18	XLD	GSD2B 300257	Xq24	<b>LAMP2</b> 309060	Lysosomal-associated membrane protein 2	Nishino et al (2000) Musumeci et al (2005)	
Myopathy with excessive autophagy	5.19	XLR	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	5.20	AR	CLN3 204200	16p12.1	<b>CLN3</b> 607042	Ceroid-lipofuscinosis, neuronal 3 (= Battenin)		
Oculopharyngeal muscular dystrophy	5.21	AD	OPMD 164300	14q11.2	<b>PABPN1</b> 602279	Polyadenylate-binding protein, nuclear 1	Brais et al (1995, 1998) Robinson et al (2005)	
Oculopharyngeal muscular dystrophy 2	5.22	AD	OPMD2 620460	7p15.2	<b>HNRNPA2B1</b> 600124	Heterogeneous nuclear ribonucleoprotein A2/B1	Kim et al (2022)	Allelic to IBMPPFD2 (group 5)
Oculopharyngodistal myopathy 1	5.23	AD	OPDM1 164310	8q22.3	<b>LRP12</b> 618299	Low density lipoprotein receptor-related protein 12	Ishiura et al (2019)	Allelic to ALS28 (group 12)
Oculopharyngodistal myopathy 2	5.24	AD	OPDM2 618940	19p13.12	<b>GIPC1</b> 605072	GIPC PDZ Domain-containing family, member 1	Deng et al (2020)	
Oculopharyngodistal myopathy 3	5.25	AD	OPDM3 619473	1q21.2	<b>NOTCH2NLC</b> ( <i>NBPF19</i> ) 618025	Notch2 N-terminal-like protein	Ogasawara et al (2020) Yu et al (2021)	Allelic to Neuronal intranuclear inclusion diseases (group 14) and CMT2 related to NOTCH2NLC (group 14)
Oculopharyngodistal myopathy 4	5.26	AD	OPDM4 619790	12q24.31	<b>RILPL1</b> 614092	Rab-interacting lysosomal protein-like 1	Yu et al (2022) Zeng et al (2022)	
Oculopharyngodistal myopathy 5	5.27	AD	OPDM5	1p21.3	<b>ABCD3</b> 170995	ATP-binding cassette, subfamily D, member 3	Cortese et al (2024)	
Oculopharyngeal myopathy with leukoencephalopathy 1	5.28	AD	OPML1 618637	10q22.3	<b>NUTM2B-ASI</b> 618639	NUTM2B antisens RNA 1	Ishiura et al (2019)	
Epidermolysis bullosa simplex associated with muscular dystrophy	5.29	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 <i>PLEC</i> (group 11)
Muscle hypertrophy	5.30	AR	MSLHP 614160	2q32.2	<b>MSTN</b> 601788	Myostatin	Schuelke et al (2004)	
Fibrodysplasia ossificans progressiva	5.31	AD	FOP 135100	2q24.1	<b>ACVR1</b> 102576	Activin A receptor, type 1	Shore et al (2006)	
Creatine phosphokinase, elevated serum (formerly HyperCKemia, idiopathic)	5.32	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	5.33	XLR	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	5.34	XLD	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)
Reducing body myopathy (1A and 1B)	5.35	XLD	RBMX1A 300717 RBMX1B 300718	Xq26.3	<b>FHL1</b> 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	5.36	XLR	EMWX 300211	Xp22.3	?	?	Ryan et al (1999)	
Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia 1	5.37	AD	IBMPPFD1 167320	9p13.3	<b>VCP</b> 601023	Valosin-containing protein	Watts et al (2004) Haubenberger et al (2005)	allelic to scapuloperoneal myopathy (group 1), IBMPPFD1 (group 4), ALS14 (group 12) and CMT2Y (group 14)
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2	5.38	AD	IBMPPFD2 615422	7p15.2	<b>HNRNPA2B1</b> 600124	Heterogeneous nuclear ribonucleoprotein A2/B1	Kim et al (2013)	Allelic to OPMD related to <i>HNRNPA2B1</i> (group 5)
Myopathy with lactic acidosis, hereditary	5.39	AR	HML 255125	12q23.3	<b>ISCU</b> 611911	Iron-sulfur cluster scaffold homolog (E. coli)	Mochel et al (2008)	

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Late onset axial myopathy related to <i>RYR1</i>	5.40	AD		19q13.2	<b><i>RYR1</i></b> 180901	Ryanodine receptor 1 (skeletal)	Løseth et al (2013)	Allelic to Congenital muscular dystrophy related to RYR1 (group 2), CMYO1A, CMYO1B, CCD, CFTD, CNMDU1, Centronuclear myopathy related to RYR1, DuCD related to RYR1 (group 3), Fetal akinesia deformation sequence related to RYR1 (group 16)
Tubular aggregate myopathy 1	5.41	AD	TAM1 160565	11p15.4	<b><i>STIM1</i></b> 605921	Stromal interaction molecule 1	Bohm et al (2013) Hedberg et al (2014) Nesin et al (2014)	Allelic to muscle weakness with hyperlaxity, orofacial abnormalities and respiratory infections (group 5)
Tubular aggregate myopathy 2	5.42	AD	TAM2 615883	12q24.31	<b><i>ORAI1</i></b> 610277	ORAI calcium release-activated calcium modulator 1	Nesin et al (2014)	
Vacuolar myopathy with CASQ1 aggregates	5.43	AD	VMCQA 616231	1q23.2	<b><i>CASQ1</i></b> 114250	Calsequestrin 1	Rossi et al (2014)	
Myopathy, sarcoplasmic body	5.44	AD	MYOSB 620286	22q12.3	<b><i>MB</i></b> 160000	Myoglobin	Olivé et al (2019)	
Myopathy with extrapyramidal signs	5.45	AR	MPXPS 615673	10q22.1	<b><i>MICU1</i></b> 605084	Mitochondrial Calcium Uptake Protein 1	Logan et al (2014)	
Muscle weakness with hyperlaxity, orofacial abnormalities and respiratory infections	5.46	AR		11p15.4	<b><i>STIM1</i></b> 605921	Stromal interaction molecule 1	Salvi et al (2021)	Allelic to TAM1 (group 5)
Wieacker-Wolff syndrome	5.47	XLR	WRWF 314580	Xq11.2	<b><i>ZC4H2</i></b> 300897	Zinc Finger C4H2 domain-containing protein	Hennekam et al (1991) Hirata et al (2013)	
Intellectual disability and myopathy syndrome	5.48	AR	IDMYS 619719	12p12.1	<b><i>ABCC9</i></b> 601439	ATP-binding cassette, subfamily C, member 9	Smeland et al (2019)	
Childhood-onset myopathy with hyperCKaemia	5.49	AR		11p11.2	<b><i>PACSIN3</i></b> 606513	Protein kinase C and casein kinase substrate in neuron 3	Distelmaier et al (2024)	

**GROUP 6. MYOTONIC SYNDROMES**

Myotonic dystrophy 1 (Steinert)	6.1	AD	DM1 160900	19q13.32	<b><i>DMPK</i></b> 605377	Dystrophia myotonica protein kinase	Renwick et al (1971) Friedrich et al (1987) Harley et al (1992) Buxton et al (1992) Aslanidis et al (1992) Mahadevan et al (1992) Fu et al (1992) Brook et al (1992)	
Myotonic dystrophy type 2 (proximal myotonic myopathy)	6.2	AD	DM2 (PROMM) 602668	3q21.3	<b><i>CNBP</i></b> (formerly <i>ZNF9</i> ) 116955	CCHC-type Zinc finger nucleic acid-binding protein (Zinc finger protein 9)	Ranum et al (1998) Liquori et al (2001)	
Myotonia, dominant (Thomsen)	6.3	AD	see under Ion channel muscle diseases (group 7)					
Myotonia, recessive (Becker)	6.4	AR	see under Ion channel muscle diseases (group 7)					
Rippling muscle disease, dominant	6.5	AD	RMD1 600332	1q41	?	?	Stephan et al (1994)	
Rippling muscle disease, dominant	6.6	AD	RMD2 606072	3p25.3	<b><i>CAV3</i></b> 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	<b><i>CAV3</i></b> 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	<b><i>HSPG2</i></b> 14246	Heparan sulfate proteoglycan of basement membrane (perlecan)	Nicole et al (1995, 2000)	
Brody disease	6.9	AR	BROD 601003	16p11.2	<b><i>ATP2A1</i></b> (formerly <i>SERCA1</i> ) 108730	ATPase, Ca <sup>++</sup> transporting, fast twitch 1	Odermatt et al (1996)	

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<b>Group 7. ION CHANNEL MUSCLE DISEASES</b>								
<i>Chloride channel</i>								
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)
<i>Sodium channel</i>								
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 CMYO22A, CMYO22B (group 3), HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16)
Hypokalaemic periodic Paralysis, type 2	7.4	AD	HOKPP2 613345	17q23.3	<i>SCN4A</i> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Bulman et al (1999) Jurkat-Rott et al (2000)	allelic to CMYO22A, CMYO22B (group 3), HYPP, PMC and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16)
Hypokalaemic periodic paralysis	7.5	AD		1q23.2	<i>ATPIA2</i> 182340	ATPase, NA+/K+ transporting alpha-2 polypeptide	Sampedro et al (2018)	
Paramyotonia congenita	7.6	AD	PMC 168300	17q23.3	<i>SCN4A</i> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Ptáček et al (1991-1993) Ebers et al (1991) Koch et al (1992) Mc Clatchey et al (1992)	allelic to CMYO22A, CMYO22B (group 3), HYPP, HOKPP2 and K-aggravated myotonia (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16)
Potassium-aggravated myotonia	7.7	AD	608390	17q23.3	<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 CMYO22A, CMYO22B (group 3), HYPP, HOKPP2 and PMC (group 7), CMS16 (group 11), Severe foetal hypokinesia related to SCN4A (group 16)
<i>Calcium channel</i>								
Hypokalaemic periodic paralysis, type 1	7.8	AD	HOKPP1 170400	1q32.1	<i>CACNA1S</i> 114208	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Fontaine et al (1994) Ptáček et al (1994) Jurkat-Rott et al (1994) Elbaz et al (1995)	Allelic to LGMD related to CACNA1S (group 1), congenital myopathy with ophthalmoplegia, CMYO18 (group 3), and MHS5 (group 8)
Acetazolamide responsive hereditary paroxysmal cerebellar ataxia (also listed in group 13 "Ataxias")	7.9	AD	APCA 108500	19p13.13	<i>CACNA1A</i> 601011	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	von Brederlow et al (1995) Vahedi et al (1995)	allelic to EA2 (group 7), SCA6 (group 13), APCA and EA2 and familial hemiplegic migraine (group 13)
Episodic ataxia type-2	7.10	AD	EA2 108500	19p13.13	<i>CACNA1A</i> 601011	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	Ophoff et al (1996) Jodice et al (1997)	allelic to APCA (group 7 and 13), EA2 and familial hemiplegic migraine (group 13), SCA6 (group 13)
<i>Potassium channel</i>								
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 hypokalaemic periodic paralysis	7.13		TTPP2 613239	17p11.2	<i>KCNJ18</i> 613236	Potassium channel, inwardly rectifying, subfamily J, member 18 (Kir2.6)	Ryan et al (2010)	
Periodic paralysis, potassium sensitive cardiodysrhythmic (Andersen's syndrome)	7.14	see LQ7 under hereditary cardiomyopathies (group 10B, online only)						
Long QT syndromes	7.15	see under hereditary cardiomyopathies (group 10B, online only)						

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<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)	allelic to CMD related to <i>RYR1</i> (group 2) CFTD, CNM related to <i>RYR1</i> , CCD, CNMDU1, minicore myopathy with external ophthalmoplegia, DuCD (group 3), late onset axial myopathy (group 5), Fetal akinesia related to <i>RYR1</i> (group 16)
Malignant hyperthermia	8.2	AD	MHS2 154275	17q11.2-q24	?	?	Levitt et al. (1992) Moslehi et al (1998)	
Malignant hyperthermia	8.3	AD	MHS3 154276	7q21-q22	?	?	Iles et al (1994)	
Malignant hyperthermia	8.4	AD	MHS4 600467	3q13.1	?	?	Sudbrak et al (1995)	
Malignant hyperthermia	8.5	AD	MHS5 601887	1q32.1	<b><i>CACNA1S</i></b> <i>114208</i>	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Monnier et al (1997)	Allelic to LGMD related to <i>CACNA1S</i> (group 1), congenital myopathy with ophthalmoplegia, CMYO18 (group 3), and HOKPP1 (group 7)
Malignant hyperthermia	8.6	AD	MHS6 601888	5p	?	?	Robinson et al (1997)	
<b>GROUP 9. METABOLIC MYOPATHIES</b>								
<i>Glycogen storage diseases</i>								
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) Sheng et al (1996)	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-glucanotransferase (glycogen debrancher enzyme)		
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) Bruno et al (2004)	
Glycogen storage disease type V (McArdle)	9.4	AR	GSD5 232600	11q13.1	<b><i>PYGM</i></b> <i>608455</i>	Glycogen phosphorylase, muscle	Mommaerts et al (1959) Schmidt et al (1959) Lebo et al (1984) Tsujino et al (1993) Tarui et al (1965)	
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	XLR	GSD9D 300559	Xq13.1	<b><i>PHKA1</i></b> <i>311870</i>	Phosphorylase kinase, alpha-1 subunit	Wehner et al (1994) Burwinkel et al (2004)	
Glycogen storage disease type XV	9.7	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	9.8	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	9.9	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	9.10	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)	
Polyglucosan Body Myopathy 2	9.11	AR	PGBM2 616199	3q24	<b><i>GYGI</i></b> <i>603942</i>	Glycogenin 1	Malfatti et al (2014)	allelic to GSD15 (group 9)
Myopathy with myalgia, increased serum creatine kinase and with or without episodic rhabdomyolysis	9.12	AR	MMCKR 620138	6p12.1	<b><i>MLIP</i></b> <i>614106</i>	Muscular LMNA-interacting protein	Abath Neto et al (2021)	
Congenital disorder of glycosylation, type Ie	9.13	AR	CDG1E 608799	20q13.13	<b><i>DPM1</i></b> <i>603503</i>	Dolichyl-phosphate mannosyltransferase 1, catalytic subunit	Kim et al (2000) Yang et al (2013)	
Congenital disorder of glycosylation, type If	9.14	AR	CDG1F 609180	17p13.1	<b><i>MPDU1</i></b> <i>604041</i>	Mannose-P-dolichol utilization defect 1	Schenk et al (2001)	
Congenital disorder of glycosylation, type It Formerly Glycogen storage disease type XIV	9.15	AR	CDG1T 614921	1p31.3	<b><i>PGM1</i></b> <i>171900</i>	Phosphoglucomutase 1	Stojkovic et al (2009)	

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Congenital disorder of glycosylation, type Iu	9.16	AR	CDG1U 615042	9q34.11	<b>DPM2</b> 603564	Dolichyl-phosphate mannosyltransferase polypeptide 2, regulatory subunit	Barone et al (2012)	
<b>Glycolytic pathway</b>								
Phosphoglycerate kinase 1 deficiency	9.17	XLR	300653	Xq21.1	<b>PGK1</b> 311800	Phosphoglycerate kinase 1	DiMauro et al (1981, 1983) Rosa et al (1982)	
Glycogen storage disease type X	9.18	AR	GSD10 261670	7p13	<b>PGAM2</b> 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	9.19	AR	GSD11 612933	11p15.1	<b>LDHA</b> 150000	Lactate dehydrogenase A	Kanno et al (1980) Scrable et al (1990)	
Glycogen storage disease type XIII	9.20	AD	GSD13 612932	17p13.2	<b>ENO3</b> 131370	Enolase 3	Comi et al (2001)	
Erythrocyte lactate transporter defect (Myopathy due to actate transporter defect)	9.21	AD	245340	1p13.2	<b>SLC16A1</b> 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	9.22	AD, AR	255110	1p32.3	<b>CPT2</b> 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	9.23	AR	CDSP 212140	5q31.1	<b>SLC22A5</b> 603377	Solute carrier family 22 (organic cation transporter), member 5	Nezu et al (1999)	
Carnitine/acyl-carnitine translocase deficiency	9.24	AR	CACTD 212138	3p21.31	<b>SLC25A20</b> 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)	9.25	AR	MADD 231680	15q24.2-q24.3	<b>ETFA</b> 608053	Electron-transfer-flavoprotein, alpha polypeptide	Indo et al (1991) Freneaux et al (1992)	
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIB)	9.26	AR	MADD 231680	19q13.41	<b>ETFB</b> 130410	Electron-transfer-flavoprotein, beta polypeptide	Colombo et al (1994)	
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIC, riboflavin responsive)	9.27	AR	MADD 231680	4q32.1	<b>ETFDH</b> 231675	Electron-transfer-flavoprotein dehydrogenase	Beard et al (1993)	
Acyl-CoA dehydrogenase (very long chain) deficiency	9.28	AR	ACADVL 201475	17p13.1	<b>ACADVL</b> 609575	Acyl-Coenzyme A dehydrogenase, very long chain	Aoyama (1993, 1995) Strauss et al (1995) Mathur et al 1999)	
Mitochondrial complex I deficiency, nuclear type 20	9.29	AR	MC1DN20 611126	3q21.3	<b>ACAD9</b> 611103	Acyl-CoA dehydrogenase family member 9 (M)	Fragaki et al (2016)	
Triglyceride storage disease with ichthyosis (Chanarin-Dorfman syndrome)	9.30	AR	CDS 275630	3p21.33	<b>ABHD5</b> 604780	Abhydrolase domain containing 5	Lefevre et al (2001)	
Neutral lipid storage disease with myopathy without ichthyosis	9.31	AR	NLSDM 610717	11p15.5	<b>PNPLA2</b> 609059	Patatin-like phosphorylase domain-containing protein 2 (Adipose triglyceride lipase)	Fischer et al (2007)	
Myoglobinuria, acute recurrent, autosomal recessive	9.32	AR	268200	2p25.1	<b>LPINI</b> 605518	lipin 1 (phosphatidic acid phosphatase 1)	Zeharia et al (2008)	
Mitochondrial myopathy with lactic acidosis	9.33	AR	MMLA 251950	7q31.1	<b>PNPLA8</b> 612123	Patatin-like phospholipase domain containing 8 (M)	Saunders et al (2015)	
Lipid storage myopathy due to Flavin adenine dinucleotide synthetase deficiency	9.34	AR	LSMFLAD 255100	1q21.3	<b>FLADI</b> 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	<b>MYH7</b> 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, CMYO7A, CMYO7B, MSM with eccentric core disease (group 3), MPD1 (group 4), CMD1S and LVNC5 (group 10A)
Familial hypertrophic cardiomyopathy, 2	10.2	AD	CMH2 115195	1q32.1	<b>TNNT2</b> 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	<b>TPM1</b> 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	<b>MYBPC3</b> 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	<b>PRKAG2</b> 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	<b>TNNI3</b> 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	<b>MYL3</b> 160790	Myosin, light chain 3, alkali; ventricular, skeletal, slow	Poetter et al (1996)	
Familial hypertrophic cardiomyopathy, 9	10.8	AD	CMH9 613765	2q31.2	<b>TTN</b> 188840	Titin	Satoh et al (1999)	Allelic to LGMDR10 (group 1), CNM related to TTN, MmD related to TTN and CMY05 (group 3), TMD (group 4), MFM9 (group 5), CMD1G (group 10A), LCCS related to TTN (group 12)
Familial hypertrophic cardiomyopathy, 10	10.9	AD	CMH10 608758	12q24.11	<b>MYL2</b> 160781	Myosin, light chain 2, regulatory, cardiac, slow	Poetter et al (1996)	Allelic to MFM12 (group 3)
Familial hypertrophic cardiomyopathy, 11	10.10	AD	CMH11 612098	15q14	<b>ACTC1</b> 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	<b>CSRP3</b> 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	<b>TNNC1</b> 191040	Slow troponin C	Landstrom et al (2008)	allelic to CMD1Z (group 10A)
Familial hypertrophic cardiomyopathy, 14	10.13	AD	CMH14 613251	14q11.2	<b>MYH6</b> 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	<b>VCL</b> 193065	Vinculin	Vasile et al (2006)	allelic to CMD1W (group 10)
Familial hypertrophic cardiomyopathy, 1	10.15	AD, digenic	CMH1 192600	20q11.21	<b>MYLK2</b> 606566	Myosin light chain kinase 2	Davis et al (2001)	
Familial hypertrophic cardiomyopathy, 1	10.16	AD	CMH1 192600	3p25.3	<b>CAV3</b> 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	<b>MYOZ2</b> 605602	Myozenin 2 (calsarcin 1)	Osio et al (2007)	
Hypertrophic cardiomyopathy, 17	10.18	AD	CMH17 613873	20q13.12	<b>JPH2</b> 605267	Junctophilin-2	Landstrom et al (2007) Matsuhita et al (2007)	
Hypertrophic cardiomyopathy, 18	10.19	AD	CMH18 613874	6q22.31	<b>PLN</b> 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	<b>CALR3</b> 611414	Calreticulin 3	Chiu et al (2007)	
Hypertrophic cardiomyopathy, 20	10.21	AD	CMH20 613876	1p31.1	<b>NEXN</b> 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	<b>ANKRD1</b> 609599	Ankyrin repeat domain-protein 1	Arimura et al (2009)	allelic to dilated cardiomyopathy (group 10A)
Hypertrophic cardiomyopathy, 21	10.23	AD	CMH21 614676	7p12.1-q21	?	?	Song et al (2006)	

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Hypertrophic cardiomyopathy, 22	10.24	AD	CMH22 615248	10q21.3	<b>MYPN</b> 608517	Myopalladin	Purevjav et al (2012)	allelic to NEM11 (group 3), CMD1KK and RCM4 (group 10A)
Hypertrophic cardiomyopathy, 23	10.25	AD	CMH23 612158	1q43	<b>ACTN2</b> 102573	Actinin alpha-2	Chiu C et al (2010)	allelic to CMYO8 (group 3), MPD6, distal myopathy with facial weakness (group 4), CMD1AA (group 10A)
Hypertrophic cardiomyopathy, 24	10.26	AD	CMH24 601493	10q23.2	<b>LDB3</b> 605906	LIM domain binding-3	Theis et al (2006)	allelic to Late onset distal myopathy (group 4), MFM4 (group 5), CMD1C and LVNC3 (group 10)
Hypertrophic cardiomyopathy, 25	10.27	AD	CMH25 607487	17q12	<b>TCAP</b> 604488	Titin-cap (telethonin)	Hayashi et al (2004)	allelic to LGMDR7 (group 1), CMD related to TCAP (group 2), CMD1N (group 10A)
Hypertrophic cardiomyopathy, 26	10.28	AD	CMH26 617047	7q32.1	<b>FLNC</b> 102565	Filamin C	Valdes-Mas et al (2014)	allelic to MFM5 (group 5) and MPD4 (group 4) and RCM5 (group 10A)
Hypertrophic cardiomyopathy, 27	10.29	AR	CMH27 618052	15q25.3	<b>ALPK3</b> 617608	Alpha Kinase 3	Almomani et al (2016)	
Hypertrophic cardiomyopathy, 28	10.30	AD	CMH28 619402	18q12.2	<b>FHOD3</b> 609691	Formin homology-2 domain-containing protein 3	Ochoa et al (2018)	
Mitochondrial complex I deficiency, nuclear type 11	10.31	AR	MC1DN11 618234	15q15.1	<b>NDUF4F1</b> 606934	NADH dehydrogenase (ubiquinone) complex I, Assembly factor 1 (M)	Fassone et al (2011)	
Combined oxydative phosphorylation deficiency 3	10.32	AR	COXPD3 610505	12q14.1	<b>TFSM</b> 604723	Ts translation elongation factor, mitochondrial (M)	Smeitink et al.(2006)	
Combined oxydative phosphorylation deficiency 8	10.33	AR	COXPD8 614096	6p21.1	<b>AARS2</b> 612035	Alanyl-tRNA synthetase 2, mitochondrial (M)	Götz et al (2011)	
Combined oxydative phosphorylation deficiency 9	10.34	AR	COXPD9 614582	3q21.1	<b>MRPL3</b> 607118	Mitochondrial ribosomal protein L3 (M)	Galmiche et al (2011)	
Combined oxydative phosphorylation deficiency 10	10.35	AR	COXPD10 614702	6q13	<b>MTO1</b> 614667	Mitochondrial tRNA translation optimization 1 (M)	Ghezzi et al (2012) Baruffini et al (2013)	
Combined oxydative phosphorylation deficiency 16	10.36	AR	COXPD16 615395	2q36.1	<b>MRPL44</b> 611849	Mitochondrial ribosomal protein L44 (M)	Carroll et al (2013)	
Mitochondrial complex IV deficiency, nuclear type 2	10.37	AR	MC4DN2 (CEMCOX1) 604377	22q13.33	<b>SCO2</b> 604272	Cytochrome c oxidase assembly protein (M)	Papadopoulou et al (1999)	allelic to CMT2 related to SCO2 (group 14)
Mitochondrial complex IV deficiency, nuclear type 6	10.38	AR	MC4DN6 (CEMCOX2) 615119	10q24.2	<b>COX15</b> 603646	Cytochrome c oxidase assembly factor Cox15 (M)	Antonicka et al (2003)	
<b>Dilated cardiomyopathies</b>								
Dilated cardiomyopathy, 1A	10.39	AD	CMD1A 115200	1q22	<b>LMNA</b> 150330	Lamin A/C	Fatkin et al (1999)	Allelic to EDMD2, EDMD3 (group1), MDCL (group 2), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
Dilated cardiomyopathy, 1B	10.40	AD	CMD1B 600884	9q13	?	?	Krajinovic et al (1995)	
Dilated cardiomyopathy, 1C, with or without LVNC	10.41	AD	CMD1C 601493	10q23.2	<b>LDB3</b> 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	10.42	AD	CMD1D 601494	1q32.1	<b>TNNT2</b> 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	10.43	AD	CMD1E 601154	3p22.2	<b>SCN5A</b> 600163	Sodium channel, voltage-gated, type V, alpha	McNair et al (2004)	allelic to BRGDA1, ATFB10, LQT3, SSS1 (group 10B)
Dilated cardiomyopathy, 1G	10.44	AD	CMD1G 604145	2q31.2	<b>TTN</b> 188840	Titin	Siu et al (1999) Gerull et al (2002) Itoh-Satoh et al (2002)	Allelic to LGMDR10 (group 1), CNM related to TTN, MmD related to TTN and CMYO5 (group 3), TMD (group 4), MFM9 (group 5), CMH9 (group 10A), LCCS related to TTN (group 12)

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Dilated cardiomyopathy, 1H	10.45	AD	CMD1H 604288	2q14-q22	?	?	Jung et al (1999)	
Dilated cardiomyopathy, 1I	10.46	AD	CMD1I 604765	2q35	<b>DES</b> 125660	Desmin	Li et al (1999)	Allelic to formerly-LGMD1-related to DES and formerly-LGM2R (group 1); MFM1 with or without ARCV7 (group 5), ARVC7 (group 10B) and myopathy, cardiomyopathy and CMS (group 11)
Dilated cardiomyopathy, 1J	10.47	AD	CMD1J 605362	6q23.2	<b>EYA4</b> 603550	Eyes absent homolog	Schönberger et al (2005)	
Dilated cardiomyopathy, 1K	10.48	AD	CMD1K 605582	6q12-q16	?	?	Sylvius et al (2001)	
Dilated cardiomyopathy, 1L	10.49	AD	CMD1L 606685	5q33.2-q33.3	<b>SGCD</b> 601411	Sarcoglycan, delta	Tsubata et al (2000)	allelic to LGMDR6 (group 1)
Dilated cardiomyopathy, 1M	10.50	AD	CMD1M 607482	11p15.1	<b>CSRP3</b> 600824	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Knoll et al (2002)	allelic to CMH12 (group 10A)
Dilated cardiomyopathy, 1N	10.51	AD	CMD1N	17q12	<b>TCAP</b> 604488	Titin-cap (Telethonin)	Knoll et al (2002)	allelic to LGMDR7 (group 1), CMD related to TCAP (group 2), CMH25 (group 10A)
Dilated cardiomyopathy, 1O	10.52	AD	CMD1O 605569	12p12.1	<b>ABCC9</b> 601439	ATP-binding cassette, subfamily C, member 9	Bienengraeber et al (2004)	allelic to ATFB12 (group 10B)
Dilated cardiomyopathy, 1P	10.53	AD	CMD1P 609909	6q22.31	<b>PLN</b> 172405	phospholamban	Schmitt et al (2003) Haghighi et al (2003, 2006)	allelic to CMH18 (group 10A)
Dilated cardiomyopathy, 1Q	10.54	AD	CMD1Q 609915	7q22.3-q31.1	?	?	Schonberger et al (2005)	
Dilated cardiomyopathy, 1R	10.55	AD	CMD1R 613424	15q14	<b>ACTC1</b> 102540	actin, alpha, cardiac muscle	Olson et al (1998) Mogensen et al (1999)	allelic to CMH11, LVNC4 (group 10A)
Dilated cardiomyopathy, 1S	10.56	AD	CMD1S 613426	14q11.2	<b>MYH7</b> 160760	Myosin heavy chain 7 (beta), cardiac muscle	Kamisago et al (2000)	Allelic to CFTD, CMY07A, CMY07B, MSM with eccentric core disease (group 3), MPD1 (group 4), CMH1 and LVNC5 (group 10A)
Dilated cardiomyopathy, 1T	10.57	AD	CMD1T	12q23.1	<b>TMPO</b> 188380	Thymopoietin (lamina-associated polypeptide 2)	Taylor et al (2005)	
Dilated cardiomyopathy, 1U	10.58	AD	CMD1U 613694	14q24.2	<b>PSEN1</b> 104311	Presenilin 1	Li et al (2006)	
Dilated cardiomyopathy, 1V	10.59	AD	CMD1V 613697	1q42.13	<b>PSEN2</b> 600759	Presenilin 2	Li et al (2006)	
Dilated cardiomyopathy, 1W	10.60	AD	CMD1W 611407	10q22.2	<b>VCL</b> 193065	Vinculin	Olson et al (2002) Vasile et al (2006)	allelic to CMH15 (group 10)
Dilated cardiomyopathy related, 1X	10.61	AR	CMD1X 611615	9q31.2	<b>FKTN</b> 607440	Fukutin	Murakami et al (2006)	allelic to MDDGC4 (group 1), MDDGA4 and MDDGB4 (group 2)
Dilated cardiomyopathy, 1Y	10.62	AD	CMD1Y 611878	15q22.2	<b>TPM1</b> 191010	Tropomyosin-1	Olson et al (2010)	allelic to CMH3 and LVNC9 (group 10A)
Dilated cardiomyopathy, 1Z	10.63	AD	CMD1Z 611879	3p21.1	<b>TNNC1</b> 191040	Slow troponin C	Mogensen et al (2004)	allelic to CMH13 (group 10A)
Dilated cardiomyopathy, 1AA, with or without LVNC	10.64	AD	CMD1AA 612158	1q42-q43	<b>ACTN2</b> 102573	Actinin alpha-2	Mohapatra et al (2003)	allelic to CMY08 (group 3), MPD6 (group 4), CMH23 (group 10A)
Dilated cardiomyopathy, 1BB	10.65	AD, AR	CMD1BB 612877	18q12.1	<b>DSG2</b> 125671	Desmoglein 2	Posch et al (2008)	allelic to ARVD10 (group 10B)
Dilated cardiomyopathy, 1CC	10.66	AD	CMD1CC 613122	1p31.1	<b>NEXN</b> 613121	Nexilin F-actin binding protein	Hassel et al (2009)	allelic to CMH20 (group 10A)
Dilated cardiomyopathy, 1DD	10.67	AD	CMD1DD 613172	10q25.2	<b>RBM20</b> 613171	RNA-binding motif protein 20	Brauch et al (2009)	
Dilated cardiomyopathy, 1EE	10.68	AD	CMD1EE 613252	14q11.2	<b>MYH6</b> 160710	Myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al (2005)	allelic to CMH14 (group 10A), SSS3 (group 10B)
Dilated cardiomyopathy, 1FF	10.69	AD	CMD1FF 613286	19q13.42	<b>TNNI3</b> 191044	Cardiac troponin I	Carballo et al (2009)	allelic to CMH7, CDM2A, RCM1 (group 10A)
Dilated cardiomyopathy, 1GG	10.70	AR	CMD1GG 613642	5p15.33	<b>SDHA</b> 600857	Succinate dehydrogenase complex, subunit a, flavoprotein (M)	Levitas et al (2010)	
Dilated cardiomyopathy, 1HH	10.71	AD	CMD1HH 613881	10q26.11	<b>BAG3</b> 603883	Bcl2-associated athanogene 3	Norton et al (2011)	allelic to MFM6 (group 5) and CMT related to BAG3 (group 14)

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Dilated cardiomyopathy, III	10.72	AD	CMD1II 615184	11q23.1	<b>CRYAB</b> 123590	Alpha-B crystallin	Inagaki et al (2006)	Allelic to MFM2 (group 5) and CMT2 related to <i>CRYAB</i> (group 14)
Dilated cardiomyopathy, IJJ	10.73	AD	CMD1JJ 615235	6q21	<b>LAMA4</b> 600133	Laminin, alpha-4	Knöll et al (2007)	
Dilated cardiomyopathy, IKK	10.74	AD	CMD1KK 615248	10q21.3	<b>MYPN</b> 608517	Myopalladin	Duboscq-Bidot (2008)	allelic to NEM11 (group 3), CMH22 and RCM4 (group 10A)
Dilated cardiomyopathy, ILL	10.75	AD	CMD1LL 615373	1p36.32	<b>PRDM16</b> 605557	PR domain-containing protein 16	Arndt et al (2013)	allelic to LVNC8 (group 10A)
Dilated cardiomyopathy, IMM	10.76	AD	CMD1MM 615396	11p11.2	<b>MYBPC3</b> 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, INN	10.77	AD	CMD1NN 615916	3p25.2	<b>RAF1</b> 164760	V-Raf-1 murine leukemia viral oncogene homolog 1	Dhandapanay et al (2014)	
Dilated cardiomyopathy, IOO	10.78	AD	CMD1OO 609915	17q22	<b>VEZFI</b> 606747	Vascular endothelial zinc finger 1	Shi et al (2023)	
Dilated cardiomyopathy related to <i>ILK</i>	10.79	AD		11p15.4	<b>ILK</b> 602366	Integrin-linked kinase	Knöll et al (2007)	
Dilated cardiomyopathy related to <i>ANKRD1</i>	10.80	AD		10q23.31	<b>ANKRD1</b> 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) allelic to CMH7, CMD1FF, RCM1 (group 10A)
Dilated cardiomyopathy, 2A	10.81	AR	CMD2A 611880	19q13.42	<b>TNNI3</b> 191044	Cardiac troponin I	Carballo et al (2009)	
Dilated cardiomyopathy, 2B	10.82	AR	CMD2B 614672	7q21.2	<b>GATAD1</b> 614518	GATA zinc finger domain-containing protein 1	Theis et al (2011)	
Dilated cardiomyopathy, 2C	10.83	AR	CDMD2C 618189	1p34.2	<b>PPCS</b> 609853	Phosphopantothenoylcysteine synthetase	Iuso et al (2018)	
Dilated cardiomyopathy, 2D	10.84	AR	CMD2D 619371	16p13.3	<b>RPL3L</b> 617416	Ribosomal protein L3-like	Ganapathi et al (2020)	
Dilated cardiomyopathy, 2E	10.85	AR	CMD2E 619492	20q13.12	<b>JPH2</b> 605267	Junctophilin-2	Vasilescu et al (2018) Jones et al (2019)	Allelic to CMH17 (group 10A)
Dilated cardiomyopathy, 2F	10.86	AR	CMD2F 619747	14q32.33	<b>BAG5</b> 603885	Bag cochaperone	Hakui et al (2022)	
Dilated cardiomyopathy, 2G	10.87	AR	CMD2G 619897	7q31.32	<b>LMOD2</b> 68006	Leiomodin 2	Ahrens-Nicklas et al (2019)	
Dilated cardiomyopathy, 2I	10.89	AR	CMD2I 620462	6p22.3	<b>CAP2</b> 618385	Cyclase-associated actin cytoskeleton regulatory protein 2	Aspit et al (2019)	
Dilated cardiomyopathy, 2J	10.90	AR	CMD2J 620635	17p11.2	<b>FLII</b> 600362	FLII actin remodeling protein	Al-Hassnan et al (2020) Ruijmbek et al (2023)	
Dilated cardiomyopathy, 3A	10.91	XLR	CMD3A	Xq28	<b>TAZ</b> 300394	Tafazzin	Gedeon et al (1995)	allelic to BTHS (group 10A)
Dilated cardiomyopathy, 3B	10.92	XLR	CMD3B 302045	Xp21.2-p21.1	<b>DMD</b> 300377	Dystrophin	Muntoni et al (1993) Milasin et al (1996)	allelic to DMD (group 1)
Dilated cardiomyopathy related to <i>SYNE1</i>	10.93	AD		6q25.2	<b>SYNE1</b> 608441	Spectrin repeat containing, nuclear envelope protein 1 (nesprin-1)	Puckelwartz et al (2010)	allelic to EDMD4 (group 1) SCAR8 (group 13), AMCM (group 16)
Dilated cardiomyopathy related to <i>CAVIN4</i>	10.94	AD		9q31.1	<b>CAVIN4</b> 617714	Caveolae-associated protein 4 (MURC)	Rodriguez et al (2011)	
Dilated cardiomyopathy related to <i>DOLK</i>	10.95	AR	CDGM1 610768	9q34.11	<b>DOLK</b> 610746	Dolichol kinase	Kranz et al (2007) Lefebvre et al (2011)	
<b>Restrictive cardiomyopathies</b>								
Restrictive cardiomyopathy, 1	10.96	AD	RCM1 115210	19q13.42	<b>TNNI3</b> 191044	Cardiac troponin I	Mogensen et al (2003)	allelic to CMH7, CMD1FF, CMD2A (group 10A)
Restrictive cardiomyopathy, 2	10.97		RCM2 609578	10q23.3	?	?	Zhang et al (2005)	
Restrictive cardiomyopathy, 3	10.98	AD	RCM3 612422	1q32.1	<b>TNNT2</b> 191045	Cardiac Troponin T	Peddy et al (2006)	allelic to CMH2, CMD1D and LVNC6 (group 10a)
Restrictive cardiomyopathy, 4	10.99	AD	RCM4 615248	10q21.3	<b>MYPN</b> 608517	Myopalladin	Purejav et al (2012)	allelic to NEM11 (group 3), CMH22 and CMD1KK (group 10A)
Restrictive cardiomyopathy, 5	10.100	AD	RCM5 617047	7q32.1	<b>FLNC</b> 102565	Filamin C	Brodehl et al (2016)	allelic to MFM5 (group 5), MPD4 (group 4) and CMH26 (group 10A)
Restrictive cardiomyopathy, 6	10.101	AR	RCM6 619433	5q31.2	<b>KIF20A</b> 605664	Kinesin family member 20A	Louw et al (2018)	

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<b>Other non arrhythmogenic hereditary cardiomyopathies</b>								
Pompe disease, Glycogenosis, generalized, cardiac form ( <i>also listed in group 9</i> )	10.102	AR	GSD2 232300	17q25.3	<b>GAA</b> 606800	Glucosidase, alpha; acid	Hers (1963) Martiniuk et al (1990) Wokke et al (1995)	Allelic to Pompe disease (formerly LGMD2V) (group 1)
Barth syndrome	10.103	XLR	BTBS 302060	Xq28	<b>TAZ</b> 300394	Tafazzin	Bolhuis et al (1991) Bione et al (1996)	allelic to CMD3A (group 10A)
Left ventricular noncompaction, 1	10.104	AD	LVNC1 604169	18q12.1	<b>DTNA</b> 601239	Dystrobrevin, alpha	Ichida et al (2001)	
Left ventricular noncompaction, 2	10.105		LVNC2 609470	11q15	?	?	Sasse-Klaassen et al (2004)	
Left ventricular noncompaction, 3	10.106	AD	LVNC3 601493	10q23.2	<b>LDB3</b> 605906	LIM domain binding-3	Vatta et al (2003)	allelic to Late onset distal myopathy (group 4), MF4 (group 5), CMH24 and CMD1C (group 10A)
Left ventricular noncompaction, 4	10.107	AD	LVNC4 613424	15q14	<b>ACTC1</b> 102540	Alpha-cardiac actin	Monserrat et al (2007)	allelic to CMH11, CMD1R (group 10A)
Left ventricular noncompaction, 5	10.108	AD	LVNC5 613426	14q11.2	<b>MYH7</b> 160760	Myosin heavy chain 7 (beta), cardiac muscle	Klaassen et al (2008)	Allelic to CFTD, CMY07A, CMY07B, MSM with eccentric core disease (group 3), MPD1 (group 4), CMH1, CMD1S (group 10A)
Left ventricular noncompaction, 6	10.109	AD	LVNC6 601494	1q32.1	<b>TNNT2</b> 191045	Cardiac Troponin T	Luedde et al (2010)	allelic to CMH2, CMD1D and RCM3 (group 10A)
Left ventricular noncompaction, 7	10.110	AD	LVNC7 615092	18q11.2	<b>MIB1</b> 608677	Mindbomb, drosophila, homolog of	Luxan et al. (2013)	
Left ventricular noncompaction, 8	10.111	AD	LVNC8 615373	1p36.32	<b>PRDM16</b> 605557	PR domain-containing protein 16	Arndt et al. (2013)	allelic to CMD1LL (group 10A)
Left ventricular noncompaction, 9	10.112	AD	LVNC9 611878	15q22.2	<b>TPMI</b> 191010	Tropomyosin-1	Probst et al. (2011)	allelic to CMH3, CMD1Y (group 10A)
Left ventricular noncompaction, 10	10.113	AD	LVNC10 615396	11p11.2	<b>MYBPC3</b> 600958	Cardiac myosin binding protein-C	Probst et al. (2011)	allelic to congenital skeletal myopathy and fatal cardiomyopathy (group 3), CMH4 and CMD1MM (group 10A)
Cardiovalvular dysplasia, X-linked	10.114	XLR	CVDPX 314400	Xq28	<b>FLNA</b> 300017	Filamin A, alpha (actin binding protein 280)	Kyndt et al (1998) Kyndt et al (2007)	
<b>GROUP 11. CONGENITAL MYASTHENIC SYNDROMES</b>								
Myasthenic syndrome, congenital, 1A, Slow-channel	11.1	AD	CMS1A 601462	2q31.1	<b>CHRNA1</b> 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	<b>CHRNBI</b> 100710	Cholinergic receptor, nicotinic, beta 1	Engel et al (1996b) Gomez et al (1996)	allelic to CMS2B, CMS2C (group 11)
Myasthenic syndrome, congenital, 3A, Slow-channel	11.3	AD	CMS3A 616321	2q37.1	<b>CHRNA1</b> 100720	Cholinergic receptor, nicotinic, delta	Gomez et al (2002)	allelic to CMS3B, CMS3C (group 11)
Myasthenic syndrome, congenital, 4A, Slow-channel	11.4	AD, AR	CMS4A 605809	17p13.2	<b>CHRNA1</b> 100725	Cholinergic receptor, nicotinic, epsilon	Ohno et al (1995) Gomez et al (1995) Engel et al (1996) Croxen et al (2002)	allelic to CMS4B, CMS4C (group 11)
Myasthenic syndrome, congenital, 1B, Fast-channel	11.5	AR	CMS1B 608930	2q31.1	<b>CHRNA1</b> 100690	Cholinergic receptor, nicotinic, alpha 1	Wang et al (1999) Shen et al (2003)	allelic to CMS1A (group 11)
Myasthenic syndrome, congenital, 2B, Fast-channel	11.6	AR	CMS2B	17p13.1	<b>CHRNBI</b> 100710	Cholinergic receptor, nicotinic, beta 1	Shen et al (2020)	allelic to CMS2A, CMS2C (group 11)
Myasthenic syndrome, congenital, 3B, Fast-channel	11.7	AR	CMS3B 616322	2q37.1	<b>CHRNA1</b> 100720	Cholinergic receptor, nicotinic, delta	Brownlow et al (2001)	allelic to CMS3A, CMS3C (group 11)
Myasthenic syndrome, congenital, 4B, Fast-channel	11.8	AR	CMS4B 616324	17p13.2	<b>CHRNA1</b> 100725	Cholinergic receptor, nicotinic, epsilon	Ohno et al (1996)	allelic to CMS4A, CMS4C (group 11)
Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	11.9	AR	CMS2C 616314	17p13.1	<b>CHRNBI</b> 100710	Cholinergic receptor, nicotinic, beta 1	Quiram et al (1999)	allelic to CMS2A, CMS2B (group 11)

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Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	11.10	AR	CMS3C 616323	2q37.1	<b>CHRNA3</b> 100720	Cholinergic receptor, nicotinic, delta	Shen et al (2002)	allelic to CMS3A, CMS3B (group 11)
Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	11.11	AR	CMS4C 608931	17p13.2	<b>CHRNA4</b> 100725	Cholinergic receptor, nicotinic, epsilon	Engel et al (1996) Ohno et al (1997)	allelic to CMS4A, CMS4B (group 11)
Myasthenic syndrome, congenital, 5	11.12	AR	CMS5 603034	3p25.1	<b>COLQ</b> 603033	Collagenic tail of endplate acetylcholinesterase	Donger et al (1998) Ohno et al (1998-2000)	
Myasthenic syndrome, congenital, 6, presynaptic	11.13	AR	CMS6 254210	10q11.23	<b>CHAT</b> 118490	Choline acetyltransferase	Ohno et al (2001) Maselli et al (2003)	
Myasthenic syndrome, congenital, 7, presynaptic and distal motor neuropathy, autosomal dominant	11.14	AD	CMS7A 616040	1q32.1	<b>SYT2</b> 600104	Synaptotagmin 2	Herrmann et al (2014)	Allelic to CMS7B (group 11) and to Distal motor neuropathy related to SYT2 (group 12)
Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive	11.15	AR	CMS7B 619461	1q32.1	<b>SYT2</b> 600104	Synaptotagmin 2	Masseli et al (2020)	Allelic to CMS7 (group 11) and to Distal motor neuropathy related to SYT2 (group 12)
Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	11.16	AR	CMS8 615120	1p36.33	<b>AGRN</b> 103320	Agrin	Huzé et al (2009)	Allelic to FADS related to AGRN (group 16)
Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	11.17	AR	CMS9 616325	9q31.3	<b>MUSK</b> 601296	Muscle-specific receptor tyrosine kinase	Chevessier et al (2004)	allelic to FADS (group 16)
Myasthenic syndrome, congenital, 10	11.18	AR	CMS10 254300	4p16.3	<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	11.19	AR	CMS11 616326	11p11.2	<b>RAPSN</b> 601592	Receptor-associated protein of the synapse, 43kD (Rapsyn)	Ohno et al (2002) Ohno et al (2003) Dunne et al (2003)	allelic to FADS (group 16)
Myasthenia, congenital, 12, with tubular aggregates	11.20	AR	CMS12 610542	2p13.3	<b>GFPT1</b> 138292	Glutamine:fructose-6-phosphate amido transferase 1	Senderek et al (2011)	
Myasthenic syndrome, congenital, 13, with tubular aggregates	11.21	AR	CMS13 614750	11q23.3	<b>DPAGT1</b> 191350	Dolichyl-phosphate N-acetylglucosamine phosphotransferase 1	Belaya et al (2012)	
Myasthenic syndrome, congenital, 14, with tubular aggregates	11.22	AR	CMS14 616228	9q22.33	<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	11.23	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	11.24	AR	CMS16 614198	17q23.3	<b>SCN4A</b> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Tsujino et al (2003)	allelic to CMYO22A, CMYO22B (group 3), HOKPP2, HYPP, PMC and K-aggravated myotonia (group 7), Severe foetal hypokinesia related to SCN4A (group 16)
Myasthenic syndrome, congenital, 17	11.25	AR	CMS17 616304	11p11.2	<b>LRP4</b> 604270	LDL receptor-related protein 4	Ohkawara et al (2014)	
Myasthenic syndrome, congenital, 18	11.26	AD	CMS18 616330	20p12.2	<b>SNAP25</b> 600322	Synaptosomal associated protein 25	Shen et al (2014)	
Myasthenic syndrome, congenital, 19	11.27	AR	CMS19 616720	10q22.1	<b>COL13A1</b> 120350	Collagen type XIII alpha 1 chain	Logan et al (2015)	
Myasthenic syndrome, congenital, 20, presynaptic	11.28	AR	CMS20 617143	2q12.3	<b>SLC5A7</b> 608761	Solute carrier family 5 (choline transporter) member 7	Bauche et al (2016)	allelic to HMND7 (group 12)
Myasthenic syndrome, congenital, 21, presynaptic	11.29	AR	CMS21 617239	10q11.23	<b>SLC18A3</b> 600336	Solute carrier family 18 (vesicular acetylcholine), member 3	O'Grady et al (2016)	
Myasthenic syndrome, congenital, 22	11.30	AR	CMS22 616224	2p21	<b>PREPL</b> 609557	Prolyl endopeptidase-like	Regal et al (2014)	

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Myasthenic syndrome, congenital, 23, presynaptic	11.31	AR	CMS23 618197	22q11.21	<b>SLC25A1</b> <i>190315</i>	Solute carrier family 25 (mitochondrial carrier, citrate transporter), member 1	Chaouch et al (2014)	
Myasthenic syndrome, congenital, 24, presynaptic	11.32	AR	CMS24 618198	15q23	<b>MYO9A</b> <i>604875</i>	Myosin IXA	O'Connor et al (2016)	
Myasthenic syndrome, congenital, 25	11.33	AR	CMS25 618323	12p13.31	<b>VAMPI</b> <i>185880</i>	Vesicle-associated membrane protein 1	Shen et al (2017)	allelic to SPAX1 (group 15)
Congenital myasthenic syndrome with nephrotic syndrome	11.34	AR	NPHS5 614199	3p21.31	<b>LAMB2</b> <i>150325</i>	Laminin-beta 2	Salpieto et al (2017) Maselli et al (2009)	
Escobar syndrome (multiple pterygium syndrome)	11.35	AR	EVMP5 265000	2q37.1	<b>CHRNA3</b> <i>100730</i>	Cholinergic receptor, nicotinic, gamma	Hoffman et al (2006) Morgan et al (2006)	
Myasthenic syndrome, with plectin defect	11.36	AR		8q24.3	<b>PLEC</b> <i>601282</i>	Plectin	Banwell et al (1999) Forrest et al (2010) Selcen et al (2011)	allelic to LGMDR17 (group 1), LGMD with ophtalmoplegia (group 1) and EBSMD (group 5)
Congenital myasthenic syndrome related to GMPPB	11.37	AR	MDDGC14 615352	3q21.31	<b>GMPPB</b> <i>615320</i>	GDP-mannose pyrophosphorylase B	Belaya et al (2015)	allelic to LGMDR19/MDDGC14 (group 1); MDDGA14 and MDDGB14 (group 2)
Presynaptic congenital myasthenic syndrome	11.38	AR		20q13.33	<b>LAMA5</b> <i>601033</i>	Laminin, alpha-5	Maselli et al (2017)	
Presynaptic congenital myasthenic syndrome related to <i>MUNC13-1</i>	11.39	AR		19p13.11	<b>UNC13A</b> <i>609894</i>	C. Elegans, homolog of UNC13A (MUNC13)	Engel et al (2016)	
Congenital myasthenic syndrome related to <i>RPH3A</i> , presynaptic	11.40	AR		12q24.13	<b>RPH3A</b> <i>612159</i>	Rabphilin 3A	Maselli et al (2018)	
Congenital myasthenic syndrome	11.41	AR		1q25.2	<b>TOR1AIP1</b> (= <i>LAP1B</i> ) <i>614512</i>	Torsin A interacting protein 1 (=Lamin Associated Peptide 1B)	Cossins et al (2020) Malfatti et al (2021)	allelic to MRRSDC (group 1)
Congenital myasthenic syndrome	11.42	AD		14q11.2	<b>CHD8</b> <i>610528</i>	Chromodomain Helicase DNA-Binding Protein 8	Lee et al (2020)	
Myasthenic syndrome, congenital associated with acetylcholine receptor deficiency	11.43	AR		2q31.1	<b>CHRNA1</b> <i>100690</i>	Cholinergic receptor, nicotinic, alpha 1	Abath Neto et al (2017)	allelic to CMS1A and CMS1B (group 11)
Myopathy, cardiomyopathy and congenital myasthenic syndrome	11.44	AR		2q35	<b>DES</b> <i>125660</i>	Desmin	Durmus et al (2016)	allelic to MFM1 with or without ARCV7 (group 5), CMD11 (group 10A), ARVC7 (group 10B)
PURA syndrome with neuromuscular junction manifestations	11.45	AD	NEDRIHF 616158	5q31.3	<b>PURA</b> <i>600473</i>	Purine-rich element-binding protein A	Qashqari et al (2022)	

**GROUP 12. SPINAL MUSCULAR ATROPHIES MOTONEURON DISEASES***Spinal muscular atrophy related to SMN1*

Spinal muscular atrophy, type I (Werdnig-Hoffman)	12.1	AR	SMA1 253300	5q13.2	<b>SMN1</b> <i>600354</i>	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	<b>SMN1</b> <i>600354</i>	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	<b>SMN1</b> <i>600354</i>	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	<b>SMN1</b> <i>600354</i>	Survival of motor neuron 1	Brahe et al (1995) Clermont et al (1995)	allelic to SMA2, SMA3, SMA1 (group 12)

*Distal spinal muscular atrophy, recessive*

Neuronopathy, distal hereditary motor, autosomal recessive 1	12.5	AR	HMNR1 604320	11q13.3	<b>IGHMBP2</b> <i>600502</i>	Immunoglobulin Mu-binding protein 2	Grohmann et al (1999, 2001)	allelic to CMT2S (group 14)
Neuronopathy, distal hereditary motor, autosomal recessive 2	12.6	AR	HMNR2 605726	9p13.3	<b>SIGMAR1</b> <i>601978</i>	Sigma non-opioid intracellular receptor 1	Christodoulou et al (2000) Li et al (2015)	allelic to ALS16 (group 12)

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Neuronopathy, distal hereditary motor, autosomal recessive 3	12.7	AR	HMNR3 607088	11q13	?	?	Viollet et al (2004)	
Neuronopathy, distal hereditary motor, autosomal recessive 4	12.8	AR	HMNR4 611067	1p36.31	<b>PLEKHG5</b> 611101	Pleckstrin homology domain and RhoGEF domain-containing protein G5	Maystadt et al (2006, 2007)	allelic to CMTRIC (group 14)
Neuronopathy, distal hereditary motor, autosomal recessive 5	12.9	AR	HMNR5 614881	2q35	<b>DNAJB2</b> 604139	DnaJ/Hsp40 homolog, subfamily B, member 2	Blumen et al (2012)	Allelic to CMT related to DNAJB2 (group 14)
Neuronopathy, distal hereditary motor, autosomal recessive 6	12.10	AR	HMNR6 620011	2p11.2	<b>REEPI</b> 609139	Receptor expression-enhancing protein 1	Schottmann et al (2015)	Allelic to HMND12 (group 12) and SPG31 (group 15)
Neuronopathy, hereditary motor, autosomal recessive 7	12.11	AR	HMNR7 619216	1p36.33	<b>VWAI</b> 611901	Von Willebrand factor A domain-containing protein 1	Deschauer et al (2021) Pagnamenta et al (2021)	
Neuronopathy, hereditary motor, autosomal recessive 8	12.12	AR	HMNR8 618912	15q21.1	<b>SORD</b> 182500	Sorbitol dehydrogenase	Cortese et al (2020)	Allelic to SORDD (group 14)
Neuronopathy, distal hereditary motor, autosomal recessive 9	12.13	AR	HMNR9 620402	16q12.3	<b>COQ7</b> 601683	Coenzyme Q7	Jacquier et al (2023) Liu et al (2023) Smith et al (2023)	Allelic to COQ10D8 (group 16)
Neuronopathy, distal hereditary motor, autosomal recessive 10	12.14	AR	HMNR10 620542	14q32.2	<b>VRK1</b>	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)
Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity	12.15	AR	HMNR11 620854	19q13.32	<b>RTN2</b> 603183	Reticulon 2	Maroofian et al (2024)	Allelic to SPEG12 (group 15)
Spinal muscular atrophy with congenital bone fractures 1	12.16	AR	SMABF1 616866	15q22.31	<b>TRIP4</b> 604501	Thyroid hormone receptor interactor 4	Knierim et al (2016)	allelic to MDCDC (group 2)
Spinal muscular atrophy with congenital bone fractures 2	12.17	AR	SMABF2 616867	10q22.1	<b>ASCCI</b> 614215	Activating signal cointegrator 1 complex subunit 1	Knierim et al (2016)	allelic to Arthrogyrosis related to ASCCI (group 16)
Spinal muscular atrophy with progressive myoclonic epilepsy	12.18	AR	SMAPME 159950	8p22	<b>ASAH1</b> 613468	N-acylsphingosine amidohydrolase 1	Zhou et al (2012)	allelic to FRBRL (#228000)
Spinal muscular atrophy and cerebellar hypoplasia	12.19	AR	PCH1C 616081	13q13.3	<b>EXOSC8</b> 606019	Exosome component 8	Boczonadi et al (2014)	
Pontocerebellar hypoplasia, type 1D	12.20	AR	PCH1D 618065	4q27	<b>EXOSC9</b> 606180	Exosome component 9	Burns et al (2018)	
Distal hereditary motor neuropathy	12.21	AR		5q23.3	<b>HINT1</b> 601314	Histidine triad nucleotide-binding protein	Zhao et al (2014)	Allelic to NMNAN (group 14)
Spinal Muscular Atrophy type IV related to CAPN1	12.22	AR		11q13.1	<b>CAPN1</b> 114220	Calpain 1	Perez-Siles et al (2022)	Allelic to SPG76 (group 15)
<b>Distal spinal muscular atrophy, dominant</b>								
Neuronopathy, distal hereditary motor, type 1	12.23	AD	HMND1 182960	7q34-q36	?	?	Gopinath et al (2007)	
Neuronopathy, distal hereditary motor, autosomal dominant 2	12.24	AD	HMND2 158590	12q24.23	<b>HSPB8</b> 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, autosomal dominant 3	12.25	AD	HMND3 608634	7q11.23	<b>HSPB1</b> 602195	Heat-shock 27-kD protein 1	Evgrafov et al (2004)	allelic to CMT2F (group 14)
Neuronopathy, distal hereditary motor, autosomal dominant 4	12.26	AD	HMND4 613376	5q11.2	<b>HSPB3</b> 604624	Heat shock 27-kD protein 3	Kolb et al. (2010)	
Neuronopathy, distal hereditary motor, autosomal dominant 5	12.27	AD	HMND5 600794	7p14.3	<b>GARS1</b> 600287	Glycyl-tRNA synthetase	Christodoulou et al (1995) Antonellis et al (2003)	allelic to CMT2D (group 14)
Neuronopathy, distal hereditary motor, autosomal dominant 6	12.28	AD	HMND6 615575	5q32	<b>FBXO38</b> 608533	F-box only protein 38	Sumner et al (2013)	

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Neuropathy, distal hereditary motor, autosomal dominant 7	12.29	AD	HMND7 158580	2q12.3	<i>SLC5A7</i> 608761	Solute carrier family 5 (choline cotransporter), member 7	McEntagart et al. (2001) Barwick et al (2012)	allelic to CMS20 (group 11)
Neuropathy, distal hereditary motor, autosomal dominant 8	12.30	AD	HMND8 600175	12q24.11	<i>TRPV4</i> 605427	Transient receptor potential cation channel, subfamily V, member 4	van der Vleuten et al (1998) Auer-Grumbach et al (2010) Deng et al (2010)	allelic to SPSMA (group 12), HSMN2C (group 14)
Neuropathy, distal hereditary motor, autosomal dominant 9 (formerly type IX)	12.31	AD	HMND9 61772	14q32.2	<i>WARS1</i> 191050	Tryptophanyl-tRNA synthetase 1	Tsai et al (2017)	
Neuropathy, distal hereditary motor, autosomal dominant 10	12.32	AD	HMND10 620080	2p23.3	<i>EMILIN1</i> 130660	Elastin microfibril interfacer 1	Capuano et al (2016)	
Neuropathy, distal hereditary motor, autosomal dominant 11	12.33	AD	HMND11 620528	9q34.11	<i>SPTAN1</i> 182810	Spectrin, alpha, nonerythrocytic 1	Beijer et al (2019)	Allelic to SPG91 (group 15)
Neuropathy, distal hereditary motor, autosomal dominant 12	12.34	AD	HMND12 614751	2p11.2	<i>REEP1</i> 609139	Receptor expression-enhancing protein 1	Beetz et al (2012)	Allelic to HMNR6 (group 12) and SPG31 (group 15)
Neuropathy, distal hereditary motor, autosomal dominant 13	12.35	AD	HMND13 619112	11q12.3	<i>BSCL2</i> 606158	Seipin	Windpassinger et al (2004)	allelic to SPG17 (group 15)
Neuropathy, distal hereditary motor, autosomal dominant 14	12.36	AD	HMND14 607641	2p13.1	<i>DCTN1</i> 601143	Dynactin 1	Puls et al (2003)	allelic to ALS related to DCTN1 (group 12)
Dominant distal hereditary motor neuropathy	12.37	AD	dHMN	16q22.1	<i>AARS1</i> 601065	Alanyl-tRNA synthetase 1	Zhao et al (2012)	allelic to CMT2N (group 14)
Hereditary motor and sensory neuropathy V	12.38	AD	HMSN5 600361	4q34.3-q35.2	?	?	Muglia et al (2008)	
Scapuloperoneal spinal muscular atrophy	12.39	AD	SPSMA 181405	12q.24	<i>TRPV4</i> 605427	Transient receptor potential cation channel, subfamily V, member 4	Isozumi et al (1996) Deng et al (2010)	allelic to HMND8 (group 12), HSMN2C (group 14)
Spinal motor neuropathy	12.40			11q23.2	<i>RBM7</i> 612413	RNA-binding motif protein 7	Giunta et al (2016)	
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 1	12.41	AD	SMALED1 158600	14q32.31	<i>DYNC1H1</i> 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	12.42	AD	SMALED2A 615290	9q22.31	<i>BICD2</i> 609797	Bicaudal D homolog 2 (Drosophila)	Neveling et al (2013) Oates et al (2013) Peeters et al (2013)	Allelic to SMALED2B (group 12) and Arthrogryposis and BICD2-related neuromuscular disease (group 16)
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 2 B	12.43	AD	SMALED2B 618291	9q22.31	<i>BICD2</i> 609797	Bicaudal D homolog 2 (Drosophila)	Ravenscroft et al (2016)	Allelic to SMALED2A (group 12) and Arthrogryposis and BICD2-related neuromuscular disease (group 16)
Spinal muscular atrophy, late-onset, Finkel type	12.44	AD	SMAFK 182980	20q13.32	<i>VAPB</i> 605704	Vesicle-associated membrane protein-associated protein B	Nishimura et al (2004)	allelic to ALS8 (group 12)
Spinal muscular atrophy, Jokela type	12.45	AD	SMAJ 615048	22q11.23	<i>CHCHD10</i> 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	12.46	AD		1q32.1	<i>SYT2</i> 600104	Synaptotagmin 2	Montes-Chinea et al (2018)	Allelic to CMS7A, CMS7B (group 11)
Distal hereditary motor neuropathies	12.47	AD		10q24.32	<i>GBF1</i> 603698	Golgi-specific brefeldin-A resistance factor 1	Mendoza-Ferreira et al (2020)	Allelic to CMT2GG (group 14)
Sensory-motor neuropathy with oculofacial apraxia	12.48	AR		4q13.3	<i>COX18</i> 610428	Cytochrome c Oxidase Assembly Factor COX18 (M)	Mavillard et al (2024)	

**Distal spinal muscular atrophy, X-linked**

Spinal and bulbar muscular atrophy, X-linked, 1 (Kennedy disease)	12.49	XLR	SMAJ1 313200	Xq12	<i>AR</i> 313700	Androgen receptor	Fishbeck et al. (1986) La Spada et al. (1991)	
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Spinal muscular atrophy, distal, X-linked 2	12.50	XLR	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	12.51	XLR	SMAX3 300489	Xq21.1	<b>ATP7A</b> 300011	ATPase, Cu <sup>++</sup> transporting, alpha polypeptide	Takata et al (2004) Kennerson et al (2010)	
Spinal muscular atrophy with respiratory distress (SMARD)	12.52	XLR		Xq12	<b>LASIL</b> 300964	Las1-like ribosome biogenesis factor	Stembalska et al (2022)	
<b>Amyotrophic lateral sclerosis (ALS)</b>								
Amyotrophic lateral sclerosis 1 (dominant)	12.53	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)	12.54	AR	ALS1 105400	21q22.11	<b>SOD1</b> 147450	Cu/Zn superoxide dismutase	Andersen et al (1995)	
Amyotrophic lateral sclerosis 2, juvenile	12.55	AR	ALS2 205100	2q33.1	<b>ALS2</b> 606352	Alsin Rho guanine nucleotide exchange factor 2	Hentati et al. (1994) Yang et al (2001) Hadano et al (2001)	allelic to IAHS (group 15)
Amyotrophic lateral sclerosis 3	12.56	AD	ALS3 606640	18q21	?	?	Hand et al (2002)	
Amyotrophic lateral sclerosis 4, juvenile	12.57	AD	ALS4 602433	9q34.13	<b>SETX</b> 608465	Senataxin	Chance et al (1998) Chen et al (2004) Moreira et al (2004)	allelic to SCAR1 (group 13)
Amyotrophic lateral sclerosis 5, juvenile	12.58	AR	ALS5 602099	15q21.1	<b>SPG11</b> 610844	Spatacsin	Hentati et al (1998) Orlacchio et al (2010)	allelic to CMT2X (group 14)
Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	12.59	AD	ALS6 608030	16p11.2	<b>FUS</b> 137070	Fused in sarcoma	Sapp et al (2003) Abalkhail et al (2003) Kwiatkowski et al (2009) Vance et al (2009)	
Amyotrophic lateral sclerosis 7	12.60	AD	ALS7 608031	20p13	?	?	Sapp et al (2003)	
Amyotrophic lateral sclerosis 8	12.61	AD	ALS8 608627	20q13.32	<b>VAPB</b> 605704	Vesicle-associated membrane protein-associated protein B and C	Nishimura et al (2004)	allelic to SMAFK (group 12)
Amyotrophic lateral sclerosis 9	12.62	AD	ALS9 611895	14q11.2	<b>ANG</b> 105850	Angiogenin	Greenway et al (2006) Wu et al (2007)	
Amyotrophic lateral sclerosis 10, with or without frontotemporal dementia	12.63	AD	ALS10 612069	1p36.22	<b>TARDBP</b> 605078	TAR DNA-binding protein	Sreedharan et al (2008)	Allelic to late-onset distal myopathy (group 4)
Amyotrophic lateral sclerosis 11	12.64	AD	ALS11 612577	6q21	<b>FIG4</b> 609390	FIG4, <i>S. Cerevisiae</i> , homolog of	Chow et al (2009)	allelic to CMT4J (group 14)
Amyotrophic lateral sclerosis 12, with or without frontotemporal dementia	12.65	AD, AR	ALS12 613435	10p13	<b>OPTN</b> 602432	Optineurin	Maruyama et al (2010)	
Amyotrophic lateral sclerosis 13, susceptibility to	12.66	AD	ALS13 183090	12q24.12	<b>ATXN2</b> 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	12.67	AD	ALS14 613954	9p13.3	<b>VCP</b> 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	12.68	XLD	ALS15 300857	Xp11.21	<b>UBQLN2</b> 300264	Ubiquilin 2	Deng et al (2011)	
Amyotrophic lateral sclerosis 16, juvenile	12.69	AR	ALS16 614373	9p13.3	<b>SIGMAR1</b> 601978	Sigma non-opioid intracellular receptor 1	Al-Saif et al (2011)	allelic to DSMA2 (group 12)
Amyotrophic lateral sclerosis 17	12.70	AD	ALS17 614696	3p11.2	<b>CHMP2B</b> 609512	Charged multivesicular body protein 2B	Parkinson et al (2006) Cox et al (20010)	
Amyotrophic lateral sclerosis 18	12.71	AD	ALS18 614808	17p13.2	<b>PFN1</b> 176610	Profilin 1	Wu et al (2012)	
Amyotrophic lateral sclerosis 19	12.72	AD	ALS19 615515	2q34	<b>ERBB4</b> 600543	V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 4	Takahashi et al (2013)	
Amyotrophic lateral sclerosis 20	12.73	AD	ALS20 615426	12q13.13	<b>HNRNPA1</b> 164017	Heterogeneous nuclear ribonucleoprotein A1	Kim et al (2013)	allelic to IBMPFD3 (group 3) and MPD3 (group 4)

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Amyotrophic lateral sclerosis 21	12.74	AD	ALS21 606070	5q31.2	<b>MATR3</b> 164015	Matrin 3	Johnson et al (2014)	allelic to VCPDM (group 4)
Amyotrophic lateral sclerosis 22, with or without frontotemporal dementia	12.75	AD	ALS22 616208	2q35	<b>TUBA4A</b> 191110	Tubulin, alpha-4A	Smith et al (2014)	
Amyotrophic lateral sclerosis 23	12.76	AD	ALS23 617839	10q22.3	<b>ANXA11</b> 602572	Annexin A11	Smith et al (2017)	Allelic to IBMWMA (group 12)
Amyotrophic lateral sclerosis 24, susceptibility to	12.77		ALS24 617892	4q33	<b>NEKI</b> 604588	Never in mitosis gene A-related kinase 1	Brenner et al (2016) Kenna et al (2016)	
Amyotrophic lateral sclerosis 25, susceptibility to	12.78	AD	ALS25 617921	12q13.3	<b>KIF5A</b> 602821	Kinesin family member 5A	Nicolas et al. (2018)	Allelic to CMT2 related to KIF5A (group 14) and SPG10 (group 15)
Amyotrophic lateral sclerosis 26, with or without frontotemporal dementia	12.79	AD	ALS26 619133	2p13.3	<b>TIA1</b> 603518	Cytotoxic granule-associated RNA binding protein	MacKenzie et al (2017)	Allelic to WDM (group 4)
Amyotrophic lateral sclerosis 27	12.80	AD	ALS27 620285	9q22.31	<b>SPTLC1</b> 605712	Serine palmitoyl transferase long-chain base subunit 1	Mohassel et al (2021)	Allelic to HSAN1A (group 14)
Amyotrophic lateral sclerosis 28	12.81	AD	ALS28 620452	8q22.3	<b>LRP12</b> 618299	Low density lipoprotein receptor-related protein 12	Kume et al (2023)	Allelic to OPDM1 (group 5)
Amyotrophic lateral sclerosis related to NEFH, susceptibility to	12.82	AD, AR	ALS1 105400	22q12.2	<b>NEFH</b> 162230	Neurofilament, heavy polypeptide	Al-Chalabie et al (1999)	Allelic to CMT2CC (group 14)
Amyotrophic lateral sclerosis related to peripherin, susceptibility to	12.83	AD	ALS1 105400	12q13.12	<b>PRPH</b> 170710	Peripherin	Gros-Louis et al (2004) Leung et al (2004)	
Amyotrophic lateral sclerosis related to dynactin 1, susceptibility to	12.84	AD	ALS1 105400	2p13.1	<b>DCTN1</b> 601143	Dynactin 1	Munch et al (2005)	Allelic to HMND14 (group 12)
Frontotemporal dementia and/or amyotrophic lateral sclerosis 1	12.85	AD	FTDALS1 105550	9p21.2	<b>C9orf72</b> 614260	Chromosome 9 open reading frame 72	Morita et al (2006) DeJesus-Hernandez (2011)	Allelic to SBMA (group 12)
Frontotemporal dementia and/or amyotrophic lateral sclerosis 2	12.86	AD	FTDALS2 615911	22q11.23	<b>CHCHD10</b> 615903	Coiled-coil-helix-coiled-coil-helix domain containing 10 (M)	Bannwarth et al (2014)	Allelic to SMAJ (group 12), IMMD (group 16)
Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	12.87	AD	FTDALS3 66437	5q35.3	<b>SQSTM1</b> 601530	Sequestosome 1	Fecto et al (2011)	Allelic to Welander-like syndrome and DMRV (group 4)
Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	12.88	AD	FTDALS4 616439	12q14.2	<b>TBK1</b> 604834	Tank-binding kinase 1	Cirulli et al. (2015) Freischmidt et al. (2015)	
Amyotrophic lateral sclerosis	12.89	AD		14q11.2	<b>LRP10</b> 609921	Low Density Lipoprotein Receptor-Related Protein 10	Ni et al (2021)	
<b>Others</b>								
Lethal Congenital Contracture Syndrome 1	12.90	AR	LCCS1 253310	9q34.11	<b>GLE1</b> 603371	GLE1, RNA export mediator	Makela-Bengs et al (1998) Nousiainen et al. (2008)	
Lethal Congenital Contracture Syndrome 2	12.91	AR	LCCS2 607598	12q13.2	<b>ERBB3</b> 190151	V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 3	Narkis et al (2007)	
Lethal Congenital Contracture Syndrome 3	12.92	AR	LCCS3 611359	19p13.3	<b>PIP5K1C</b> 606102	Phosphatidylinositol-4-phosphate 5-kinase, type I, gamma	Narkis et al (2007)	
Lethal Congenital Contracture Syndrome 4	12.93	AR	LCCS4 614915	12q23.2	<b>MYBPC1</b> 160794	Myosin-binding protein C, slow type	Markus et al (2012)	Allelic to CMYO16 (group 3), DA1B (group 16)
Lethal Congenital Contracture Syndrome 5	12.94	AR	LCCS5 615368	19p13.2	<b>DNM2</b> 602378	Dynamamin 2	Koutsopoulos et al (2013)	Allelic to CMD related to <i>DNM2</i> (group 2), CNM1 (group 3), Distal myopathy related to <i>DNM2</i> (group 4), and CMTDIB (group 14)

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Lethal Congenital Contracture Syndrome 6	12.95	AR	LCCS6 616248	14q32.33	<b>ZBTB42</b> 613915	Zinc finger and BTB domain-containing protein 42	Patel et al (2014)	
Lethal Congenital Contracture Syndrome 7	12.96	AR	LCCS7 616286	17q21.2	<b>CNTNAP1</b> 602346	Contactin-associated protein 1	Laquerriere et al (2014) Lakhani et al (2017)	Allelic to CMT related to <b>CNTNAP1</b> (group 14)
Lethal Congenital Contracture Syndrome 8	12.97	AR	LCCS8 616287	12q13.12	<b>ADCY6</b> 600294	Adenylate cyclase 6	Laquerriere et al (2014) Agolini et al (2020)	
Lethal Congenital Contracture Syndrome 9	12.98	AR	LCCS9 616503	6q24.2	<b>ADGRG6</b> 612243	Adhesion G protein-coupled receptor G6	Ravenscroft et al (2015)	
Lethal Congenital Contracture Syndrome 10	12.99	AR	LCCS10 617022	14q24.3	<b>NEK9</b> 609798	Never in mitosis gene A-related kinase 9	Casey et al (2016)	
Lethal Congenital Contracture Syndrome 11	12.100	AR	LCCS11 617194	15q21.2	<b>GLDN</b> 608603	Gliomedin	Maluenda et al (2016)	
Lethal Congenital Contracture Syndrome related to <b>TTN</b>	12.101	AR		2q31.2	<b>TTN</b> 188840	Titin	Chervinski et al (2018)	Allelic to LGMDR10 (group 1), CNM related to <b>TTN</b> , MmD related to <b>TTN</b> and <b>CMYO5</b> (group 3), TMD (group 4), <b>MFMY9</b> (group 5), <b>CMH9</b> (group 10A), <b>CMD1G</b> (group 10A)
Spinal muscular atrophy with pontocerebellar hypoplasia, type 1b	12.102	AR	PCH1B 614678	9p13.2	<b>EXOSC3</b> 606489	Exosome component 3	Rudnik-Schöneborn et al (2013)	
Spinal muscular atrophy with or without pontocerebellar hypoplasia	12.103	AR	PCH1 607596	14q32.2	<b>VRK1</b> 602168	Vaccinia related kinase 1	Renbaum et al (2009) Stoll et al (2016)	Allelic to <b>HMNR10</b> (group 12), Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis (group 14)
Pontocerebellar hypoplasia with spinal muscular atrophy	12.104	AR	HSMN6B 616505	5q22.1	<b>SLC25A46</b> 610826	Solute Carrier Family 25, Member 46	van Dijk et al (2017)	
Brown-Vialetto-van Laere syndrome 1	12.105	AR	BVVL1 211530	20p13	<b>SLC52A3</b> 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	12.106	AR	BVVL2 614707	8q24.3	<b>SLC52A2</b> 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 <b>HEXB</b>	12.107	AR		5q13.3	<b>HEXB</b> 606873	Hexosaminidase B	Rattay et al (2013)	Allelic to Sandhoff disease, adult type (OMIM #268800)
Spinal muscular atrophy, related to <b>PRUNE1</b>	12.108	AR		1q21.3	<b>PRUNE1</b> 617413	Prune exopolyphosphatase 1	Iacomino et al (2017)	
Spinal muscular atrophy with pontocerebellar hypoplasia related to <b>KIF26B</b>	12.109	AD		1q44	<b>KIF26B</b> 614026	Kinesin family member 26B	Wojcik et al (2018)	
Peripheral neuropathy, myopathy, hoarseness and hearing loss	12.110	AD	PNMH 614369	19q13.33	<b>MYH14</b> 608568	Myosin, heavy chain 14, non muscle	Choi et al (2011)	
Lower motor neuron disease with respiratory failure related to <b>MAPT</b>	12.111	AD		17q21.31	<b>MAPT</b> 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	12.112	AR	PNRIID 618124	21q22.3	<b>MCM3AP</b> 603294	Minichromosome maintenance 3-associated protein	Schuurs-Hoeijmakers et al (2013) Ylikallio et al (2017)	Allelic to CMT2 (group 14)
Inclusion body myopathy and brain white matter abnormalities	12.113	AD	IBMWMA 619733	10q22.3	<b>ANXA11</b> 602572	Annexin A11	Leoni et al (2021)	Allelic to ALS23 (group 12)
Mitochondrial complex IV deficiency, nuclear type 11	12.114	AR	MC4DN11 619054	1q44	<b>COX20</b> 614698	Cytochrome c Oxidase Assembly Factor COX20 (M)	Dong et al (2021)	
Progressive neuropathy	12.115	AR		9q21.2	<b>PSAT1</b> 610936	Phosphoserine Aminotransferase 1	Debs et al (2021)	
Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures	12.116	AR	CONDSIAS 618170	1p34.3	<b>ADPRHL2</b> 610624	ADP-Ribosylhydrolase-Like 2	Danhauser et al (2018) Ghosh et al (2018)	
Spinobulbar muscular atrophy (SBMA)	12.117	AD		9p21.2	<b>C9orf72</b> 614260	Chromosome 9 open reading frame 72	Radziwonik et al (2022)	Allelic to FTDALS1 (group 12)

(continued on next page)

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<b>GROUP 14. HEREDITARY MOTOR SENSORY NEUROPATHIES (HMSN)</b>								
<b>A. Charcot-Marie-Tooth neuropathy, type 1 (demyelinating)</b>								
<i>Autosomal dominant (AD-CMT1)</i>								
Charcot-Marie-Tooth disease, type 1A	14.1	AD	CMT1A 118220	17p12	<b>PMP22</b> 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	<b>MPZ</b> 159440	Myelin protein zero	Bird et al (1982) Guiloff et al (1982) Hayasaka et al (1993) Kulkens et al (1993) Street et al (2002, 2003)	allelic to CMTDID, CMT2I, CMT2J, DSS and CHN (group14)
Charcot-Marie-Tooth disease, type 1C	14.3	AD	CMT1C 601098	16p13.13	<b>LITAF</b> 603795	Lipopolysaccharide-induced TNF-alpha factor	Street et al (2003)	
Charcot-Marie-Tooth disease, type 1D	14.4	AD	CMT1D 607678	10q21.3	<b>EGR2</b> 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	<b>PMP22</b> 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	<b>PMP22</b> 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	<b>NEFL</b> 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	<b>PMP2</b> 170715	Peripheral myelin protein 2	Hong et al (2016)	
Charcot-Marie Tooth disease, demyelinating type 1H	14.9	AD	CMT1H 619764	14q32.12	<b>FBLN5</b> 604580	Fibulin 5	Auer-Grumbach et al (2011)	
Charcot-Marie-Tooth neuropathy, type 1I	14.10	AD	CMT1I 619742	12q23.3	<b>POLR3B</b> 614366	Polymerase III, RNA, Subunit B	Djordjevic et al (2021) Xue et al (2021)	
Charcot-Marie-Tooth disease, demyelinating, type 1J	14.11	AD	CMT1J 620111	6q21.31	<b>ITPR3</b> 147267	Inositol 1,4,5-triphosphate receptor, type 3	Rönkkö et al (2020)	
Charcot-Marie-Tooth with congenital vertical talus	14.12	AD	CVT 192950	2q31.1	<b>HOXD10</b> 142984	Homeobox D10	Shrimpton et al (2004)	
Slowed nerve conduction velocity	14.13	AD	SNCV 608236	8p23.3	<b>ARHGEF10</b> 608136	Rho guanine-nucleotide exchange factor-10	De Jonghe et al (1999) Verhoeven et al (2003)	
<i>Dominant intermediate (CMTDI)</i>								
Charcot-Marie-Tooth disease, dominant intermediate A	14.14	AD	CMT2GG 606483	10q24.1-q25.1	<b>GBF1</b> 603698	Golgi-specific brefeldin-A resistance factor 1	Verhoeven et al (2001) Mendoza-Ferreira et al (2020)	allelic to Distal hereditary motor neuropathies (group 12)
Charcot-Marie-Tooth disease, dominant intermediate B	14.15	AD	CMTDIB 606482	19p13.2	<b>DNM2</b> 602378	Dynamin 2	Zuchner et al (2005)	allelic to CMD related to <i>DNM2</i> (group 2), <i>CNM1</i> (group 3), distal myopathy related to <i>DNM2</i> (group 4) and <i>LCCS5</i> (group 12) Allelic to <i>MLASA2</i> (group 16)
Charcot-Marie-Tooth disease, dominant intermediate C	14.16	AD	CMTDIC 608323	1p35.1	<b>YARS</b> 603623	Tyrosyl-tRNA synthetase	Jordanova et al (2003, 2006)	
Charcot-Marie-Tooth disease, dominant intermediate D	14.17	AD	CMTDID 607791	1q23.3	<b>MPZ</b> 159440	Myelin protein zero	Mastaglia et al (1999)	allelic to CMT1B, CMT2I, CMT2J, DSS and CHN (group14)
Charcot-Marie-Tooth disease, dominant intermediate E	14.18	AD	CMTDIE 614455	14q32.33	<b>INF2</b> 610982	Inverted formin 2	Boyer et al (2011)	
Charcot-Marie-Tooth disease, dominant intermediate F	14.19	AD	CMTDIF 615185	3q28.33	<b>GNB4</b> 610863	Guanine nucleotide-binding protein, beta-4	Soong et al (2013)	
Charcot-Marie-Tooth disease, dominant intermediate G	14.20	AD	CMTDIG 617882	8p21.2	<b>NEFL</b> 162280	Neurofilament, light polypeptide 68kDa	Berciano et al (2015)	allelic to CMT1F and CMT2E (group 14)
Charcot-Marie Tooth disease, intermediate	14.21	AD		1p21.2-p13.3	<b>CFAP276 (C1orf194)</b> 618682	Cilia- and flagella-associated protein 276	Sun et al (2019)	

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<b>Autosomal recessive (AR-CMT1 or CMT4)</b>								
Charcot-Marie-Tooth disease, type 4A	14.22	AR	CMT4A 214400	8q21.11	<b>GDAP1</b> 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	14.23	AR	CMT4B1 601382	11q21	<b>MTMR2</b> 603557	Myotubularin-related protein-2	Bolino et al (1996, 2000) Previtali et al (2003)	
Charcot-Marie-Tooth disease, type 4B2	14.24	AR	CMT4B2 604563	11p15.4	<b>SBF2</b> 607697	SET-binding factor 2	Azzedine et al (2003) Senderek et al (2004)	
Charcot-Marie-Tooth disease, type 4B3	14.25	AR	CMT4B3 615284	22q13.33	<b>SBF1</b> 603560	SET-binding factor 1	Nakhro et al (2013)	
Charcot-Marie-Tooth disease, type 4C	14.26	AR	CMT4C 601596	5q32	<b>SH3TC2</b> 608206	SH3 domain and tetratricopeptide repeats domain 2	LeGuern et al (1996) Senderek et al (2003)	
Charcot-Marie-Tooth disease, type 4D	14.27	AR	CMT4D 601455	8q24.22	<b>NDRG1</b> 605262	N-myc downstream regulated gene 1	Kalaydjieva et al (1996, 2000) Hunter et al (2003)	
Neuropathy, congenital hypomyelinating, 1	14.28	AR	CHN1 605253	10q21.3	<b>EGR2</b> 129010	Early growth response 2	Warner et al (1998)	allelic to CMT1D and DSS (group 14)
Neuropathy, congenital hypomyelinating, 2	14.29	AR	CHN2 605253	1q23.3	<b>MPZ</b> 159440	Myelin protein zero	Warner et al. (1996)	allelic to CMT1B, CMTDID, CMT2I, CMT2J, DSS (group 14)
Charcot-Marie-Tooth disease, type 4F	14.30	AR	CMT4F 614895	19q13.2	<b>PRX</b> 605725	Periaxin	Delague et al (2000) Guilbot et al (2001)	
Neuropathy, hereditary motor and sensory, Russe type	14.31	AR	HMSNR 605285	10q22.1	<b>HK1</b> 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	14.32	AR	CMT4H 609311	12p11.21	<b>FGD4</b> 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	14.33	AR	CMT4J 611228	6q21	<b>FIG4</b> 609390	FIG4, <i>S. Cerevisiae</i> , homolog of	Chow et al (2007)	allelic to ALS11 (group 12)
Charcot-Marie-Tooth disease, type 4K	14.34	AR	CMT4K 616684	9q34.2	<b>SURF1</b> 185620	Surfeit 1 (M)	Echaniz-Laguna et al (2013)	
Charcot-Marie-Tooth disease	14.35	AR		14q32	<b>AHNAK2</b> 608570	Ahnak nucleoprotein 2	Tey et al (2019)	
Charcot-Marie-Tooth disease related to <i>CNTNAP1</i>	14.36	AR		17q21.2	<b>CNTNAP1</b> 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	14.37	XLD	CMTX1 302800	Xq13.1	<b>GJB1</b> 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	14.38	XLR	CMTX2 302801	Xp22.2	?	?	Ionasecu et al (1992)	
Charcot-Marie-Tooth neuropathy, X-linked recessive, 3	14.39	XLR	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)	14.40	XLR	COWCK 310490	Xq26.1	<b>AIFM1</b> 300169	Apoptosis-inducing factor, mitochondria-associated, 1 (M)	Priest et al (1995) Rinaldi et al (2012)	allelic to cerebellar ataxia (group 13) and COXPD6 (group 16)
Charcot-Marie-Tooth disease, X-linked recessive, 5	14.41	XLR	CMTX5 311070	Xq22.3	<b>PRPS1</b> 311850	Phosphoribosyl pyrophosphate synthetase 1	Kim et al (2007)	
Charcot-Marie-Tooth disease, X-linked dominant, 6	14.42	XLD	CMTX6 300905	Xp22.11	<b>PDK3</b> 300906	Pyruvate dehydrogenase kinase, isoenzyme 3	Kennerson et al (2013)	
<b>Déjerine-Sottas syndrome (DSS or CMT3)</b>								
Déjerine-Sottas hypertrophic neuropathy, dominant	14.43	AD	DSS 145900	17p12	<b>PMP22</b> 601097	Peripheral myelin protein 22	Roa et al (1993)	allelic to CMT1A, CMT1E and HNPP (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.44	AD	DSS 145900	1q23.3	<b>MPZ</b> 159440	Myelin protein zero	Hayasaka et al (1993)	allelic to CMT1B, CMTDID, CMT2J, CMT2I and CHN (group 14)

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Déjerine-Sottas hypertrophic neuropathy, dominant	14.45	AD (digenic)	DSS 145900	10q21.3 and Xq13	<i>EGR2</i> 129010+ <i>GJB1</i> 304040	Early growth response 2 + Gap junction protein, beta 1	Chung et al (2005)	<i>EGR2</i> : allelic to CMT1D and CHN (group 14) <i>GJB1</i> : allelic to CMTX1 (group 14)
Déjerine-Sottas hypertrophic neuropathy, recessive	14.46	AR	DSS 145900	19q13.2	<i>PRX</i> 605725	Periaxin	Delague et al (2000) Boerkoel et al (2001)	allelic to CMT4F (group 14)
<b>B. Charcot-Marie-Tooth neuropathy, type 2 (axonal) = CMT2</b>								
<i>Autosomal dominant CMT2</i>								
Charcot-Marie-Tooth disease, type 2A1	14.47	AD	CMT2A1 118210	1p36.22	<i>KIF1B</i> 605995	Kinesin family member 1B	Zhao et al (2001)	
Charcot-Marie-Tooth disease, axonal, type 2A2A	14.48	AD	CMT2A2A 609260	1p36.22	<i>MFN2</i> 608507	Mitofusin 2	Ben Othmane et al (1993) Züchner et al (2004)	allelic to CMT2A2B and HMSN6A (group14)
Charcot-Marie-Tooth disease, type 2B	14.49	AD	CMT2B 600882	3q21.3	<i>RAB7</i> 602298	RAS-associated protein RAB7	Kwon et al (1995) Pericak-Vance et al (1997) Kok et al (2003)	
Hereditary motor and sensory neuropathy, type IIC	14.50	AD	HMSN2C 606071	12q24.11	<i>TRPV4</i> 605427	Transient receptor potential cation channel, subfamily V, member 4	Klein et al (2003) McEntagart et al (2005) Auer-Grumbach et al (2010) Deng et al (2010) Landouere et al (2010)	allelic to HMND8 and SPSMA (group 12)
Charcot-Marie-Tooth disease, type 2D	14.51	AD	CMT2D 601472	7p14.3	<i>GARS1</i> 600287	Glycyl-tRNA synthetase	Ionasescu et al (1996) Antonellis et al (2003)	allelic to HMND5 (group12)
Charcot-Marie-Tooth disease, type 2E	14.52	AD	CMT2E 607684	8p21.2	<i>NEFL</i> 162280	Neurofilament, light polypeptide	Birouk et al (2003) Claramunt et al (2005)	allelic to CMT1F, CMTDIG (group 14)
Charcot-Marie-Tooth disease, type 2F	14.53	AD, AR	CMT2F 606595	7q11.23	<i>HSPB1</i> 602195	Heat-shock 27-kD protein 1	Ismailov et al (2001) Evgrafov et al (2004) Abati et al (2021)	allelic to HMND3 (group 14)
Charcot-Marie-Tooth disease, type 2H	14.54	AD	CMT2H 607731	8q13-q23	?	?	Barhoumi et al (2001)	maybe allelic to CMT4A (group14)
Charcot-Marie-Tooth disease, type 2I	14.55	AD	CMT2I 607677	1q23.3	<i>MPZ</i> 159440	Myelin protein zero	Auer-Grumbach et al (2003)	allelic to CMT1B, CMTDID, CMT2J, DSS, CHN (group 14)
Charcot-Marie-Tooth disease, type 2J	14.56	AD	CMT2J 607736	1q23.3	<i>MPZ</i> 159440	Myelin protein zero	De Jonghe et al (1999) Chapon et al (1999)	allelic to CMT1B, CMTDID, CMT2I, DSS, CHN (group 14)
Charcot-Marie-Tooth disease, type 2K	14.57	AD, AR	CMT2K 607831	8q21.11	<i>GDAP1</i> 606598	Ganglioside-induced differentiation-associated protein 1	Nelis et al (2002) Birouk et al (2003) Claramunt et al (2005)	allelic to CMT4A and CMTRIA (group 14)
Charcot-Marie-Tooth disease, type 2L	14.58	AD	CMT2L 608673	12q24.23	<i>HSPB8</i> 608014	Heat-shock 22-kD protein 8	Tang et al (2004, 2005)	allelic to Rimmed vacuole myopathy and distal myopathy and motor neuropathy (group 4) and HMND2 (group 12)
Charcot-Marie-Tooth disease, type 2N	14.59	AD	CMT2N 613287	16q22.1	<i>AARS1</i> 601065	Alanyl-tRNA synthetase 1	Latour et al (2010)	allelic to dHMN (group 12)
Charcot-Marie-Tooth disease, type 2O	14.60	AD	CMT2O 614228	14q32.31	<i>DYNC1H1</i> 600112	Dynein, cytoplasmic 1, heavy chain 1	Weedon et al (2011)	allelic to SMALED1 (group 12)
Charcot-Marie-Tooth disease, type 2P	14.61	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	14.62	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	14.63	AD	CMT2U 616280	12q13.3	<i>MARS1</i> 156560	Methionyl-tRNA synthetase	Gonzalez et al (2013)	Allelic to SPG70 (group 15)
Charcot-Marie-Tooth disease, type 2V	14.64	AD	CMT2V 616491	17q21.2	<i>NAGLU</i> 609701	N-acetyl-alpha-glucosaminidase	Tetreault et al (2015)	
Charcot-Marie-Tooth disease, type 2W	14.65	AD	CMT2W 616625	5q31.3	<i>HARS</i> 142810	Histidyl-tRNA synthetase	Vester et al (2013) Safka-Brozkova et al (2015)	
Charcot-Marie-Tooth disease, type 2Y	14.66	AD	CMT2Y 616687	9p13.3	<i>VCP</i> 601023	Valosin-containing protein	Gonsalez et al (2014)	allelic to Scapuloperoreal myopathy (group 1), IBMPFD1 (groups 4 and 5), ALS14 (group 12)
Charcot-Marie-Tooth disease, type 2Z	14.67	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	14.68	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	14.69	AD	CMT2DD 618036	1p13.1	<i>ATPIA1</i> 182310	ATPase, Na <sup>+</sup> /K <sup>+</sup> transporting, alpha-1 polypeptide	Lassuthova et al (2018)	

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Charcot-Marie-Tooth disease, axonal, type 2FF	14.70	AD	CMT2FF 619519	1q23.2	<b>CADM3</b> 609743	Cell adhesion molecule 3	Rebelo et al (2021)	
Hereditary motor and sensory neuropathy, Okinawa type	14.71	AD	HMSNO 604484	3q12.2	<b>TFG</b> 602498	TRK-fused gene	Takehima 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	14.72	AD	HMSN6A 601152	1p36.22	<b>MFN2</b> 608507	Mitofusin 2	Zuchner et al. (2006)	Allelic to CMT2A2A and CMT2A2B (group 14)
CMT2 related to <i>KIF5A</i>	14.73	AD		12q13.3	<b>KIF5A</b> 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	14.74	AD	CMT2	1q13.5	<b>DGAT2</b> 606983	Diacylglycerol O-acyltransferase 2	Hong et al (2016)	
Charcot-Marie-Tooth disease, axonal, related to <i>BAG3</i>	14.75	AD		10q26.11	<b>BAG3</b> 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>	14.76	AD	CMT2	20p12.2	<b>JAG1</b> 601920	Jagged 1	Sullivan et al (2020)	
Charcot-Marie-Tooth disease, axonal, related to <i>NOTCH2NLC</i>	14.77	AD	CMT2	1q21.2	<b>NOTCH2NLC</b> ( <i>NBPF19</i> ) 618025	Notch2 N-terminal-like protein	Liao et al (2021)	Allelic to OPDM3 (group 5) and Neuronal intranuclear inclusion disease (group 14)
Charcot-Marie-Tooth disease, axonal, related to <i>CRYAB</i>	14.78	AD	CMT2	11q23.1	<b>CRYAB</b> 123590	Alpha-B crystallin	Cortese et al (2023)	Allelic to MFM2 (group 5) and CMDIII (group 10A)
Charcot-Marie-Tooth disease, axonal, related to <i>NARS1</i>	14.79	AD	CMT2	18q21.31	<b>NARS1</b> 108410	Asparaginyl-tRNA Synthetase 1	Beijer et al (2024)	Allelic to NEDMILG (group 14)
<b>Autosomal recessive CMT2</b>								
Charcot-Marie-Tooth disease, axonal, type 2A2B	14.80	AR	CMT2A2B 617087	1p36.22	<b>MFN2</b> 608507	Mitofusin 2	Polke et al (2011)	allelic to CMT2A2A and HMSN6A (group14)
Charcot-Marie-Tooth disease, axonal, type 2B1	14.81	AR	CMT2B1 605588	1q22	<b>LMNA</b> 150330	Lamin A/C	Bouhouch et al (1999) De Sandre et al (2002) Worman and Bonne (2007)	Allelic to EDMD2, EDMD3, 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	14.82	AR	CMT2B2 605589	19q13.33	<b>PNKP</b> 605610	Polynucleotide kinase 3-prime phosphatase	Leal et al (2009) Leal et al (2018)	
Charcot-Marie-Tooth disease, axonal, type 2EE	14.83	AR	CMT2EE 618400	2p23.3	<b>MPV17</b> 137960	MPV17, mouse, homolog of (M)	Choi et al (2015) Baumann et al (2019)	
Charcot-Marie-Tooth disease, axonal, related to <i>DNAJB2</i>	14.84	AR		2q35	<b>DNAJB2</b> 604139	DnaJ/Hsp40 homolog, subfamily B, member 2	Gess et al (2014)	allelic to DSMA5 (group 12)
Neuromyotonia and axonal neuropathy, autosomal recessive	14.85	AR	NMAN 137200	5q23.3	<b>HINT1</b> 601314	Histidine triad nucleotide binding protein 1	Zimon et al (2012)	Allelic to Distal hereditary motor neuronopathy (group 12)
Charcot-Marie-Tooth disease, recessive intermediate, A	14.86	AR	CMTRIA 608340	8q21.11	<b>GDAP1</b> 606598	Ganglioside-induced differentiation-associated protein 1	Nelis et al (2002)	allelic to CMT4A and CMT2K (group 14)
Charcot-Marie-Tooth disease, recessive intermediate, B	14.87	AR	CMTRIB 613641	16q23.1	<b>KARS</b> 601421	Lysyl-tRNA synthetase	McLaughlin et al (2010)	
Charcot-Marie-Tooth disease, recessive intermediate, C	14.88	AR	CMTRIC 615376	1p36.31	<b>PLEKHG5</b> 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	14.89	AR	CMTRID 616039	12q24.31	<b>COX6A1</b> 602072	Cytochrome c oxidase subunit 6a1 (M)	Tamiya et al (2014)	
Charcot-Marie-Tooth disease, type 2R	14.90	AR	CMT2R 615490	4q31.3	<b>TRIM2</b> 614141	Tripartite motif-containing protein 2	Ylikallio et al. (2013) Pehlivan et al (2015)	
Charcot-Marie-Tooth disease, type 2S	14.91	AR	CMT2S 616155	11q13.3	<b>IGHMBP2</b> 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	14.92	AR	CMT2T 617017	3q25.2	<b>MME</b> 120520	Membrane metalloendopeptidase	Higuchi et al (2016)	allelic to SCA43 (group 15)
Charcot-Marie-Tooth disease, type 2X	14.93	AR	CMT2X 616668	15q21.1	<b>SPG11</b> 610844	SPG11 vesicle trafficking associated Spatacsin	Montecchiani et al (2015)	allelic to ALS5 (group 12)

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Early-onset axonal Charcot-Marie-Tooth with ataxia	14.94	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	14.95	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>	14.96	AR		22q13.33	<i>SCO2</i> 604272	Cytochrome c oxidase assembly protein 2	Rebello et al (2018)	allelic to MC4DN2 (CEMCOX1) (group 10A)
Charcot-Marie-Tooth disease, axonal; related to <i>SACS</i>	14.97	AR		13q12.12	<i>SACS</i> 604490	Sacsin	Souza et al (2018)	allelic to <i>SACS</i> (group 13) and <i>SPAX6</i> (group 15)
Sorbitol dehydrogenase deficiency with peripheral neuropathy (CMT2 AR)	14.98	AR	<i>SORDD</i> 618912	15q21.1	<i>SORD</i> 182500	Sorbitol Dehydrogenase	Cortese et al (2020)	Allelic with <i>HMNR8</i> (group 15)
Charcot-Marie-Tooth disease, axonal	14.99	AR		21q22.3	<i>MCM3AP</i> 603294	Minichromosome maintenance 3-associated protein	Sedghi et al (2019)	allelic to <i>PNRIID</i> (group 12)
Charcot-Marie-Tooth disease, axonal	14.100	AR		12q13.3	<i>B4GALNT1</i> 601823	Beta-1,4-N-Acetyl-galactosaminyl transferase 1	Hong et al (2021)	allelic to <i>SPG26</i> (group 15)
Charcot-Marie-Tooth disease, axonal	14.101	AR		19p13.11	<i>MYO9B</i> 602129	Myosin IXB	Cipriani et al (2022)	
<b>C. CMT Distal = Distal hereditary motor neuropathies (dHMN) = spinal CMT or distal spinal muscular atrophy (DSMA) See under MOTOR NEURONE DISEASES (Group 12)</b>								
<b>D. Other Hereditary sensory and autonomic neuropathy (HSAN)</b>								
Hereditary sensory and autonomic neuropathy, type IA	14.102	AD	<i>HSAN1A</i> 162400	9q22.31	<i>SPTLC1</i> 605712	Serine palmitoyl transferase long-chain base subunit 1	Nicholson et al (1996) Bejaoui et al (2001) Dawkins et al (2001)	allelic to Amyotrophic lateral sclerosis (group 12)
Hereditary sensory and autonomic neuropathy, type IB with cough and gastroesophageal reflux	14.103	AD	<i>HSAN1B</i> 608088	3p24-p22	?	?	Kok et al (2004)	
Hereditary sensory and autonomic neuropathy, type IC	14.104	AD	<i>HSAN1C</i> 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	14.105	AR	<i>HSAN2A</i> 201300	12p.13.33	<i>WNK1</i> 605232	Protein kinase, lysine deficient 1	Lafreniere et al (2004) Shekarabi et al (2008)	
Hereditary sensory and autonomic neuropath, type IIB	14.106	AR	<i>HSAN2B</i> 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	14.107	AR	<i>CIP</i> 24300	2q24.3	<i>SCN9A</i> 603415	Sodium channel, voltage-gated alpha subunit	Yuan et al (2013)	allelic to <i>SFNP</i> (group 14)
Hereditary sensory and autonomic neuropathy type III	14.108	AR	<i>HSAN3</i> 223900	9q31.3	<i>ELP1</i> 603722	Inhibitor of kappa light polypeptide gene enhancer in B cells, kinase complex associated protein ( <i>IKBKAP</i> )	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	14.109	AR	<i>CIPA</i> 256800	1q23.1	<i>NTRK1</i> 191315	Neurotrophic tyrosine kinase, receptor, type 1	Indo et al (1996)	
Hereditary sensory and autonomic neuropathy type V	14.110	AR	<i>HSAN5</i> 608654	1p13.1	<i>NGF</i> 162030	Nerve growth factor (beta polypeptide)	Einarsdottir et al (2004)	
Hereditary sensory and autonomic neuropathy type VI	14.111	AR	<i>HSAN6</i> 614653	6p12.1	<i>DST</i> 113810	Dystonin	Edvardson et al (2012)	
Neuropathy, hereditary sensory and autonomic, type VII	14.112	AD	<i>HSAN7</i> 615548	3p22.2	<i>SCN11A</i> 604385	Sodium channel, voltage-gated alpha subunit	Leipold et al (2013)	allelic to <i>FEPS3</i> (group 14)
Hereditary sensory and autonomic neuropathy type VIII	14.113	AR	<i>HSAN8</i> 616488	9q34.12	<i>PRDM12</i> 616458	PR Domain-containing protein 12	Chen et al (2015)	
Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay	14.114	AR	<i>HSAN9</i> 615031	14q32.31	<i>TECPR2</i> 615000	Tectonin beta-propeller repeat containing protein 2	Oz-Levi et al (2012)	allelic to <i>SPG49</i> (group 15)
Hereditary sensory neuropathy, type ID	14.115	AD	<i>HSN1D</i> 613708	14q22.1	<i>ATL1</i> 606439	Atlantin GTPase 1	Guelly et al (2011)	allelic to <i>SPG3A</i> (group 15)

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Hereditary sensory neuropathy, type IE	14.116	AD	HSN1E 614116	19p13.2	<b>DNMT1</b> 126375	DNA methyltransferase 1	Klein et al (2011)	
Neuropathy, hereditary sensory, type IF	14.117	AD	HSN1F 615632	11q13.1	<b>ATL3</b> 609369	Atlantin GTPase 3	Kornak et al (2014)	
Hereditary sensory neuropathy, type IIC	14.118	AR	HSN2C 614213	2q37.3	<b>KIF1A</b> 601255	Kinesin family member 1A	Riviere et al (2011)	allelic to SPG30 (group 15)
Ataxia, posterior column, with retinitis pigmentosa (PCARP)	14.119	AR	AXPC1 609033	1q32.3	<b>FLVCR1</b> 609144	Feline leukemia subgroup C receptor 1	Rajadhyaksha et al (2010)	
Absence of pain, Congenital	14.120	AR		22q11.21	<b>CLTCLI</b> 601273	Clathrin, heavy polypeptide-like 1	Nahorski et al (2015)	
Marsili syndrome (insensitivity to pain, congenital, AD)	14.121	AD	MARSIS 147430	14q11.2	<b>ZFH2</b> 617828	Zinc finger homeobox 2	Habib et al (2018)	
Erythromelalgia, Primary	14.122	AD	SFNP 133020	2q24.3	<b>SCN9A</b> 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	14.123	AD	FEPS3 615552	3p22.2	<b>SCN11A</b> 604385	Sodium channel, voltage-gated alpha subunit	Zhang et al (2013) Huang et al (2014)	allelic to HSAN7 (group 14)
Polyneuropathy with erythromelalgia	14.124	AR		1q25.3	<b>NMNAT2</b> 608701	Nicotinamide nucleoside adenylyltransferase 2	Huppke et al (2019)	
Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome	14.125	AR	CANVAS 614575	4p14	<b>RF1</b> 102579	Replication Factor C Subunit 1	Cortese et al (2019)	
<b>E. Other complex neuropathy syndromes</b>								
Peripheral neuropathy and agenesis of the corpus callosum (Charlevoix disease)	14.126	AR	ACCPN 218000	15q14	<b>SLC12A6</b> ( <b>KCC3</b> ) 604878	Solute carrier family 12 (potassium/chloride transporter), member 6	Casaubon et al (1996) Howard et al (2002)	
Peripheral neuropathy and deafness, autosomal dominant	14.127	AD		1p34.3	<b>GJB3</b> 603324	Gap junction protein, beta 3	Lopez-Bigas et al (2001)	
Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy)	14.128	AD	HNA 162100	17q25.3	<b>SEPT9</b> 604061	Septin 9	Pellegrino et al (1996) Kuhlenbaumer et al (2005)	
Giant axonal neuropathy-1	14.129	AR	GAN1 256850	16q23.2	<b>GAN1</b> 605379	Gigaxonin	Ben Hamida et al (1997) Bomont et al (2000)	
Giant axonal neuropathy-2	14.130	AD	GAN2 610100	1q23.2	<b>DCAF8</b> 615820	DDB1- and CUL4-associated factor 8	Klein et al (2014)	
Congenital cataracts, facial dysmorphism and neuropathy	14.131	AR	CCFDN 604168	18p23	<b>CTDPI</b> 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	14.132	AR		14q32.2	<b>VRK1</b> 602168	Vaccinia related kinase 1	Gonzaga-Jauregui et al (2013)	Allelic to HMNR10 and PCH1 (group 12)
Neuropathy, hereditary sensory, with spastic paraplegia	14.133	AR	256840	5p15.2	<b>CCT5</b> 610150	Chaperonin containing T-complex polypeptide1, subunit 5	Bouhouche et al (2006)	
Neuronal intranuclear inclusion diseases	14.134	AD	NIID 603472	1q21.2	<b>NOTCH2NLC</b> ( <b>NBPF19</b> ) 618025	Notch2 N-terminal-like protein	Tian et al (2019) Sone et al (2019) Ishiura et al (2019)	Allelic to OPDM3 (group5) and CMT2 related to NOTCH2NLC (group 14)
Hereditary peripheral neuropathy (CMT ?)	14.135	AD		17q25.1	<b>SLC9A3R1</b> ( <b>EBP50</b> ) 604990	Solute carrier family 9, member 3, regulator 1	Song et al (2020)	
Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities	14.136	AR, AD	NEDMILG 619091	18q21.31	<b>NARS1</b> 108410	Asparaginyl-tRNA Synthetase 1	Manole et al (2020)	Allelic to CMT2 related to NARS1 (group 14)

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<b>Group 16. MITOCHONDRIAL MYOPATHIES</b>								
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1	16.1	AD, AR	PEOA1 157640 (AD) 258450 (AR)	15q26.1	<i>POLG</i> 174763	Polymerase, DNA, gamma (M)	Van Goethem et al (2001)	allelic to SANDO (group 13), PEOB1, MTDPS4A, MTDPS4B (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2	16.2	AD	PEOA2 609283	4q35	<i>SLC25A4</i> 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	16.3	AD	PEOA3 609286	10q24.31	<i>TWINK</i> 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	16.4	AD	PEOA4 610131	17q23.3	<i>POLG2</i> 604983	Polymerase DNA, gamma 2 (M)	Longley et al (2006)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5	16.5	AD	PEOA5 613077	8q22.3	<i>RRM2B</i> 604712	Ribonucleotide reductase, M2 B (M)	Tynnismaa et al (2009)	allelic to MTDPS8A and MTDPS8B (group 17)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6	16.6	AD	PEOA6 615156	10q21.3	<i>DNA2</i> 601810	DNA replication helicase 2 (M)	Ronchi et al. (2013)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 1	16.7	AR	PEOB1 258450	15q26.1	<i>POLG</i> 174763	Polymerase, DNA, gamma (M)	Deschauer et al (2007)	allelic to SANDO (group 13), PEOA1, MTDPS4A, MTDPS4B (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	16.8	AR	PEOB2 616479	2p25.3	<i>RNASEH1</i> 604123	Ribonuclease H1 (M)	Reyes et al (2015)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3	16.9	AR	PEOB3 617069	16q21	<i>TK2</i> 188250	Thymidine kinase, mitochondrial (M)	Tynnismaa et al (2012)	allelic to MTDPS2 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4	16.10	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	16.11	AR	PEOB5 618098	17p11.2	<i>TOP3A</i> 601243	DNA topoisomerase III	Nicholls et al. (2018)	
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	16.12	AR	MTDPS1 603041	22q13.33	<i>TYMP</i> 131222	Thymidine phosphorylase	Nishino et al. (1999)	
Mitochondrial DNA depletion syndrome 2 (myopathic type)	16.13	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)	16.14	AR	MTDPS3 251880	2p13.1	<i>DGUOK</i> 601465	Deoxyguanosine kinase (M)	Mandel et al. (2001)	Allelic to PEOB4 (group 16)
Mitochondrial DNA depletion syndrome 4A (Alpers type)	16.15	AR	MTDPS4A 203700	15q26.1	<i>POLG</i> 174763	Polymerase, DNA, gamma (M)	Naviaux and Nguyen (2004)	Allelic to SANDO (group 13), MTDPS4B, PEOA1, PEOB1 (group 16)

(continued on next page)

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Mitochondrial DNA depletion syndrome 4B (MNGIE type)	16.16	AR	MTDPS4B 613662	15q26.1	<b>POLG</b> 174763	Polymerase, DNA, gamma ( <b>M</b> )	Van Goethem et al. (2003)	Allelic to SANDO (group 13), MTDPS4A, PEOA1, PEOB1 (group 16)
Mitochondrial DNA depletion syndrome 11	16.17	AR	MTDPS11 615084	20p11.23	<b>MGME1</b> 615076	Mitochondrial genome maintenance exonuclease 1 ( <b>M</b> )	Kornblum et al. (2013)	
Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type)	16.18	AD	MTDPS12A 617184	4q35.1	<b>SLC25A4</b> 103220	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 ( <b>M</b> )	Thompson et al. (2016)	Allelic to PEOA2 and MTDPS12B (group 16)
Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type)	16.19	AR	MTDPS12B 615418	4q35.1	<b>SLC25A4</b> 103220	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 ( <b>M</b> )	Palmieri et al. (2005)	Allelic to PEOA2 and MTDPS12A (group 16)
Mitochondrial DNA depletion syndrome 14 (cardioencephalomyopathic type)	16.20	AR	MTDPS14 616896	3q29	<b>OPA1</b> 605290	OPA1 protein ( <b>M</b> )	Spiegel et al. (2016)	Allelic to Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia and neuropathy (group 16)
Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia and neuropathy	16.21	AD	125250	3q29	<b>OPA1</b> 605290	OPA1 protein ( <b>M</b> )	Amati-Bonneau et al (2008) Hudson et al (2008)	Allelic to MTDPS14 (group 16)
Myopathy, lactic acidosis, and sideroblastic anemia 1	16.22	AR	MLASA1 600462	12q24.33	<b>PUS1</b> 608109	Pseudourine synthase 1 ( <b>M</b> )	Bykhovskaya et al (2004) Fernandez-Vizarra (2007)	
Myopathy, lactic acidosis, and sideroblastic anemia-2	16.23	AR	MLASA2 613561	12p11.21	<b>YARS2</b> 610957	Tyrosyl-tRNA synthetase 2 ( <b>M</b> )	Riley et al (2010)	Allelic to CMTDIC (group 14)
Isolated mitochondrial myopathy	16.24	AD	IMMD 616209	22q11.23	<b>CHCHD10</b> 615903	Coiled-coil-helix-coiled-coil-helix domain containing 10 ( <b>M</b> )	Ajrroud-Driss et al (2014)	allelic to FTDALS2 and SMAJ (group 12)
Combined oxidative phosphorylation deficiency 43	16.25	AR	COXPD43 618851	17q13.3	<b>TIMM22</b> 607251	Translocase of inner mitochondrial membrane 22 ( <b>M</b> )	Pacheu-Grau et al (2018)	
Mitochondrial complex IV deficiency, nuclear type 18	16.26	AR	MC4DN18 619062	16p11.2	<b>COX6A2</b> 602009	Cytochrome c-oxidase, subunit 6A2 ( <b>M</b> )	Inoue et al (2019)	
Mitochondrial myopathy with severe neurological manifestations	16.27	AR		8q24.13	<b>TMEM65</b> 616609	Transmembrane protein 65 ( <b>M</b> )	Nasli et al (2017)	
Infantile-onset multisystem disease with progressive muscle weakness	16.28	AR	IMNEPD 616263	17q23.1	<b>PTRH2</b> 608625	Peptidyl-tRNA hydrolase 2	Hu et al (2014)	
Myopathy, mitochondrial and cerebellar ataxia (also listed in group 13)	16.29	AR	MMYAT 617675	1q22	<b>MSTO1</b> 617619	Misato 1, Mitochondrial Distribution and Morphology Regulator ( <b>M</b> )	Nasca et al (2017) Ardicli et al (2019) Donkervoort et al (2019)	allelic to CMD related to <i>MSTO1</i> (group 2)
Mitochondrial myopathy, episodic with or without atrophy and reversible leukoencephalopathy	16.30	AR	MEOAL 251900	19p13.2	<b>FDX2</b> 614585	Ferredoxin ( <b>M</b> )	Gurgel-Giannetti et al (2019)	
Mitochondrial complex I deficiency, nuclear type 29	16.31	AR	MC1DN29 618250	11q14.1	<b>TMEM126B</b> 615533	Transmembrane protein 126B ( <b>M</b> )	Alston et al (2016) Sanchez-Caballero et al (2016)	
Mitochondrial myopathy with lactic acidosis, cognitive impairment and autistic features	16.32	XL		Xp22.11	<b>APOO</b> 300753	Apolipoprotein O ( <b>M</b> )	Beninca et al (2021)	
Myopathy, mitochondrial progressive, with congenital cataract and developmental delay	16.33	AR	MPMCD 613076	16p13.3	<b>GFER</b> 600924	Growth Factor ERV1-Like	Di Fonzo et al (2009)	
Mitochondrial complex IV deficiency, nuclear type 22	16.34	AR	MC4DN22 619355	14q24.2	<b>COX16</b> 618064	Cytochrome c Oxidase Assembly Factor 16 ( <b>M</b> )	Wintjes et al (2021)	
Mitochondrial myopathy related to <i>SLC25A26</i>	16.35	AR		3p14.1	<b>SLC25A26</b> 611037	Solute carrier family 25 (mitochondrial carrier, phosphate carrier), member 26 ( <b>M</b> )	Schober et al (2022)	

(continued on next page)

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Mitochondrial myopathy and multiple mtDNA deletions	16.36	AR		9q34.11	<b>ENDOG</b> 600440	Endonuclease G, mitochondrial (M)	Nasca et al (2022)	
Multiple mitochondrial dysfunctions syndrome 10	16.37	AR	MMDS10 620960	2q11.2	<b>CIAO1</b> 604333	WD40 repeat-containing protein CIAO1	Maio et al (2024)	
<b>Group 17. OTHER NEUROMUSCULAR DISORDERS</b>								
Torsion dystonia 1, early onset	17.1	AD	DYT1 128100	9q34.11	<b>TORIA</b> 605204	Torsin 1A	Ozelius et al (1997) Ikeuchi et al (1999)	
Myoclonic dystonia 11	17.2	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)	17.3	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	17.4	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	17.5	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	17.6	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	17.7	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	17.8	AR	CFEOM5 66219	4q25	<b>COL25A1</b> 610004	Collagen, type XXV, alpha-1	Shinwari et al (2015)	
Arthrogryposis, distal, type 1A	17.9	AD	DA1A 108120	9p13.3	<b>TPM2</b> 190990	Tropomyosin-2 (beta)	Sung et al (2003)	Allelic to NEM4 and CMYP23 (group 3), DA2B4 (group 16)
Arthrogryposis, distal, type 1B	17.10	AD	DA1B 614335	12q23.2	<b>MYBPCI</b> 160794	Myosin-binding protein C, slow type	Gurnett et al (2010)	Allelic to CMYP16 (group 3), LCCS4 (group 12)
Arthrogryposis, distal, type 2A, Freeman-Sheldon syndrome	17.11	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	17.12	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	17.13	AD	DA2B2 618435	11p15.5	<b>TNNT3</b> 600692	Troponin T3, fast skeletal	Sung et al (2003)	Allelic to Nemalin myopathy with distal arthrogryposis (group 3)
Arthrogryposis, distal, type 2B3	17.14	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	17.15	AD	DA2B4 108120	9p13.3	<b>TPM2</b> 190990	Tropomyosin-2 (beta)	Tajsharghi et al (2007) Ochala et al (2007)	Allelic NEM4 and CMYP23 (group 3), DA1A (group 16)
Arthrogryposis, distal, type 3	17.16	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	17.17	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	17.18	AR	DA5D 615065	2q37.1	<b>ECEL1</b> 605896	Endothelin-converting enzyme like 1	McMillin et al (2013)	
Arthrogryposis, Distal, type 7 (Trismus-pseudocampodactyly syndrome)	17.19	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	17.20	AD	DA10 187370	2q31.3-q32.1	?	?	Stevenson et al (2006a) Stevenson et al (2006b)	
Arthrogryposis, distal type 11	17.21	AD	DA11 620019	7q31.2	<b>MET</b> 164860	MET protooncogene	Zhou et al (2019)	
Arthrogryposis, distal type 12	17.22	AR	DA12 620545	11q24.3	<b>ADAMTS15</b> 607509	A disintegrin-like and metalloproteinase with thrombospondin type 1 motif 15	Boschann et al (2022)	

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Arthrogryposis multiplex congenita with nesprin-1 defect	17.23	AR	AMCM 618484	6q25.2	<i>SYNE1</i> 608441	Spectrin repeat containing, nuclear envelope 1 (Nesprin-1)	Attali et al (2009)	allelic to EDMD4 (group 1), DCM related to <i>SYNE1</i> (group 10A) and SCAR8 (group 13)
Arthrogryposis and BICD2-related neuromuscular disease	17.24	AD		9q22.31	<i>BICD2</i> 609797	Bicaudal D homolog 2 (Drosophila)	Storbeck et al (2017)	Allelic to SMALED2A and SMALED2B (group 12)
Arthrogryposis related to <i>ASCC1</i>	17.25	AR		10q22.1	<i>ASCC1</i> 614215	Activating signal cointegrator 1 complex subunit 1	Böhm et al (2019)	allelic to SMABF2 (group 12)
Arthrogryposis multiplex congenita 7, X-linked	17.26	XLR	AMC7 301127	Xq25	<i>THOC2</i> 300395	Tho complex, subunit 2	Tamhankar et al. (2021) Dubucs et al (2023)	
Neurodevelopmental disorder with microcephaly, arthrogryposis and structural brain anomalies	17.27	AR	NEDMABA 618622	2q21.1	<i>SMPD4</i> 610457	Sphingomyelin phosphodiesterase 4, neutral membrane	Magini et al (2019)	
Fetal akinesia deformation sequence 1	17.28	AR	FADS1 208150	9q31.3	<i>MUSK</i> 601296	Muscle-specific receptor tyrosine kinase	Tan-Sindhunata et al (2015)	allelic to CMS9 (group 11)
Fetal akinesia deformation sequence 3	17.29	AR	FADS3 618389	4p16.3	<i>DOK7</i> 610285	Downstream of tyrosin kinase 7	Vogt et al (2009)	allelic to CMS10 (group 11)
Fetal akinesia deformation sequence 2	17.30	AR	FADS2 618388	11p11.2	<i>RAPSN</i> 601592	Receptor-associated protein of the synapse, 43kD (Rapsyn)	Vogt et al (2008)	allelic to CMS11 (group 11)
Fetal akinesia deformation sequence 4	17.31	AR	FADS4 618393	17p13.2	<i>NUP88</i> 602552	Nucleoporin 88kD	Bonnin et al. (2018)	
Fetal akinesia deformation sequence related to <i>RYR1</i>	17.32	AR		19q13.2	<i>RYR1</i> 180901	Ryanodine receptor	Romero et al (2003) McKie et al (2014) Alkhunaizi et al (2019)	Allelic to Congenital muscular dystrophy related to <i>RYR1</i> (group 2), <i>CMYP1A</i> , <i>CMYP1B</i> , <i>CCD</i> , <i>CFTD</i> , <i>CNMDU1</i> , Centronuclear myopathy related to <i>RYR1</i> , DuCD related to <i>RYR1</i> (group 3), Late onset axial myopathy (group 5), <i>MHS1</i> (group 8)
Fetal akinesia deformation related to <i>AGRN</i>	17.33	AR		1p36.33	<i>AGRN</i> 103320	Agrin	Geremek et al (2020)	Allelic to CMS8 (group 11)
Severe foetal hypokinesia related to <i>SCN4A</i>	17.34	AR		17q23.3	<i>SCN4A</i> 603967	Sodium channel, voltage-gated, type IV, alpha subunit	Zaharieva et al (2019)	allelic to <i>CMYP22A</i> , <i>CMYP22B</i> (group 3), <i>HYPP</i> , <i>HOKPP2</i> , <i>PMC</i> and <i>K-aggravated myotonia</i> (group 7), <i>CMS16</i> (group 11) allelic to <i>LIS9</i> (#618325)
Spectraplakopinopathy	17.35	AR		1p34.3	<i>MACF1</i> 608271	Microtubule-actin cross-linking factor 1	Kang et al (2020)	
Segmental amyoplasia with Distal Arthrogryposis	17.36	AD	DA1C 619110	16p11.2	<i>MYLFP</i> 617378	Myosin Light Chain Phosphorylatable Fast Skeletal Muscle	Chong et al (2020)	
Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis	17.37	AD	POIKTMP 615704	11q12.1	<i>FAM111B</i> 615584	Family with sequence similarity 111, member B	Mercier et al (2015)	
<b>Mitochondrial multisysteme syndromes</b>								
Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression	17.38	AR	MECREN 618416	19p13.11	<i>SLC25A42</i> 610823	Solute carrier family 25, member 42 (M)	Shamseldin et al (2015)	
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	17.39	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)	17.40	AR	MTDP8A 612075	8q22.3	<i>RRM2B</i> 604712	Ribonucleotide reductase, M2B (M)	Bourdon et al (2007)	allelic to <i>PEOA5</i> (group 16) and <i>MTDPS8B</i> (group 17)
Mitochondrial DNA depletion syndrome 8B (MNGIE type)	17.41	AR	MTDP8B 612075	8q22.3	<i>RRM2B</i> 604712	Ribonucleotide reductase, M2B (M)	Bourdon et al (2007)	allelic to <i>PEOA5</i> (group 16) and <i>MTDPS8A</i> (group 17)

(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)
Mitochondrial DNA depletion syndrome 9 (Encephalomyopathic type with methylmalonic aciduria)	17.42	AR	MTDPS9 245400	2p11.2	<b>SUCLG1</b> 611224	Succinate-CoA ligase, alpha subunit (M)	Ostergaard et al. (2007)	
Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	17.43	AR	MTDPS13 615471	6q16.1-q16.2	<b>FBXL4</b> 605654	F-box and leucine-rich repeat protein 4	Bonnen et al. (2013) Gai et al. (2013)	
Combined oxidative phosphorylation deficiency 6;	17.44	XLR	COXPD6 300816	Xq26.1	<b>AIFM1</b> 300169	Apoptosis-inducing factor, mitochondria-associated, 1 (M)	Morton et al (2017)	Allelic to cerebellar ataxia (group 13) and COCK (group 14)
Combined oxidative phosphorylation deficiency 44	17.45	AR	COXPD44 618855	2q33.3	<b>FASTKD2</b> 612322	Fast kinase domains 2 (M)	Yoo et al (2017)	
Combined oxidative phosphorylation deficiency 48	17.46	AR	COXPD48 619012	3q11.2	<b>NSUN3</b> 617491	NOP2/SUN RNA Methyltransferase Family Member 3	Van Haute et al (2016)	
Combined oxidative phosphorylation deficiency 50	17.47	AR	COXPD50 619025	3p25.1	<b>MRPS25</b> 611987	Mitochondrial ribosomal protein S25 (M)	Bugiardini et al (2019)	
Coenzyme Q10 deficiency 1	17.48	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)	17.49	AR	SCAR9 (COQ10D4) 612016	1q42.13	<b>ADCK3</b> ( <b>COQ8A</b> , <b>CABC1</b> ) 606980	Coenzyme Q8A (M)	Lagier-Tourenne et al (2008) Mollet et al (2008)	
Coenzyme Q10 deficiency 5	17.50	AR	COQ10D5 614654	16q21	<b>COQ9</b> 612837	Coenzyme Q9 (M)	Rahman et al (2001) Duncan et al (2009)	
Coenzyme Q10 deficiency 6	17.51	AR	COQ10D6 614650	14q24.3	<b>COQ6</b> 614647	Coenzyme Q6 (M)	Heeringa et al (2011)	
Coenzyme Q10 deficiency 7	17.52	AR	COQ10D7 607426	9q34.11	<b>COQ4</b> 612898	Coenzyme Q4 (M)	Brea-Calvo et al (2015)	
Coenzyme Q10 deficiency 8	17.53	AR	COQ10D8 616733	16p12.3	<b>COQ7</b> 601683	Coenzyme Q7 (M)	Freyer et al (2015)	Allelic to HMNR9 (group 12)
Mitochondrial DNA depletion syndrome 20 (MNGIE type)	17.54	AR	MTDPS20 619780	17q12	<b>LIG3</b> 600940	Ligase III DNA ATP-Dependent	Bonora et al (2021)	

**GENE TABLE 2025 NEW REFERENCES****GROUP 1. MUSCULAR DYSTROPHIES**

Iruzubieta P, Damborenea A, Ioghen M, Bajew S, Fernandez-Torron R, Topf A, et al. Biallelic variants in SNUPN cause a limb girdle muscular dystrophy with myofibrillar-like features. *Brain* awae046, 2024. PMID: 38366623 [Item #1.42]

**GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES**

Dofash LNH, Miles LB, Saito Y, Rivas E, Calcinotto V, Oveissi S, et al. HMGCS1 variants cause rigid spine syndrome amenable to mevalonic acid treatment in an animal model. *Brain*. 2024 Nov 12;awae371. doi:10.1093/brain/awae371. Epub ahead of print. PMID: 39531736 [Item #2.18]

**GROUP 3. CONGENITAL MYOPATHIES**

Johari M, Töpf A, Folland C, Duff J, Dofash L, Marti P, et al. Loss-of-function variants in JPH1 cause congenital myopathy with prominent facial and ocular involvement. *J Med Genet*. 2024 Sep 24;61(10):992-998. doi:10.1136/jmg-2024-109970. PMID: 39209426 [Item #3.66]

Warman-Chardon J, Hartley T, Marshall AE, McBride A, Couse M, Macdonald W, et al. Biallelic SOX8 Variants Associated With Novel Syndrome With Myopathy, Skeletal Deformities, Intellectual Disability, and Ovarian Dysfunction. *Neurol Genet*. 2023 Sep 19;9(5):e200088. doi:10.1212/NXG.000000000200088. PMID: 38235364 [Item #3.67]

**GROUP 4. DISTAL MYOPATHIES**

Hackman P, Rusanen SM, Johari M, Vihola A, Jonson PH, Sarparanta J, et al. Dominant distal myopathy 3 (MPD3) caused by a deletion in the HNRNPA1 gene. *Neurol. Genet*. 7: e632, 2021. PMID: 34722876 [Item #4.7]

Zibold J, Lessard LER, Picard F, da Silva LG, Zadorozhna Y, Streichenberger N, et al. The new missense G376V-TDP-43 variant induces late-onset distal myopathy but not amyotrophic lateral sclerosis. *Brain*. 2024 May 3;147(5):1768-1783. doi:10.1093/brain/awad410. PMID: 38079474 [Item #4.30]

**GROUP 5. OTHER MYOPATHIES**

Cortese A, Beecroft SJ, Facchini S, Curro R, Cabrera-Serrano M, Stevanovski I, et al. A CCG expansion in ABCD3 causes oculopharyngodistal myopathy in individuals of European ancestry. *Nat Commun*. 2024 Jul 27;15(1):6327. PMID: 39068203 doi:10.1038/s41467-024-49950-2. [Item #5.27]

Ishiura H, Shibata S, Yoshimura J, Suzuki Y, Qu W, Doi K, et al. Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy

and an overlapping disease. *Nature Genet*. 51: 1222-1232, 2019. PMID: 31332380 [Item #5.28]

Smeland MF, McClenaghan C, Roessler HI, Savelberg S, Hansen GAM, Hjeltnes H, et al. ABCC9-related intellectual disability myopathy syndrome is a K(ATP) channelopathy with loss-of-function mutations in ABCC9. *Nature Commun*. 10: 4457, 2019. PMID: 31575858 [Item #5.48]

Distelmaier F, Sezer A, Helm C, Waldmüller S, Seibt A, Gangfuß A, et al. Biallelic truncating variants in PACSIN3 cause childhood-onset myopathy with hyperCKaemia. *Brain*. 2024 Jul 5;147(7):e45-e49. doi:10.1093/brain/awae099. PMID: 38637313 [Item #5.49]

**GROUP 10A HEREDITARY CARDIOMYOPATHIES (NON-ARRHYTHMOGENIC)**

Shi H.-Y, Xie M-S, Guo Y-H, Yang C-X, Gu J-N, Qiao Q, et al. VEZF1 loss-of-function mutation underlying familial dilated cardiomyopathy. *Europ. J. Med. Genet*. 66: 104705, 2023. PMID: 36657711 [Item #10.78]

Aspit L, Levitas A, Etzion S, Krymko H, Slanovic L, Zarivach R, et al. CAP2 mutation leads to impaired actin dynamics and associates with supraventricular tachycardia and dilated cardiomyopathy. *J. Med. Genet*. 56: 228-235, 2019. PMID: 30518548 [Item #10.89]

Al-Hassnan ZN, Almesned A, Tulbah S, Alakhfash A, Alhadeq F, Alruwaili N, et al. Categorized genetic analysis in childhood-onset cardiomyopathy. *Circ. Genom. Precis. Med*. 13: 504-514, 2020. PMID: 32870709 [Item #10.90]

Ruijmbek CWB, Housley F, Idrees H, Housley MP, Pestel J, Keller L, et al. Biallelic variants in FLII cause pediatric cardiomyopathy by disrupting cardiomyocyte cell adhesion and myofibril organization. *JCI Insight* 8: e168247, 2023. PMID: 37561591 [Item #10.90]

**GROUP 11 CONGENITAL MYASTHENIC SYNDROMES**

Qashqari H, McNiven V, Gonorazky H, Mendoza-Londono R, Hassan A, Kulkarni T, et al. PURA syndrome: neuromuscular junction manifestations with potential therapeutic implications. *Neuromuscul Disord*. 2022 Oct;32(10):842-844. doi:10.1016/j.nmd.2022.09.007. Epub 2022 Sep 22. PMID: 36210261. [Item #11.45]

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

Maroofian R, Sarraf P, O'Brien TJ, Kamel M, Cakar A, Elkhateeb N, et al. Reticulon 2 deficiency results in an autosomal recessive distal motor neuropathy with lower limb spasticity. *Brain* awae091, 2024. PMID: 38527963 [Item #12.15]

Mavillard F, Guerra-Castellano A, Guerrero-Gómez D, Rivas E, Cantero G, Servian-Morilla E, et al. A splice-



altering homozygous variant in COX18 causes severe sensory-motor neuropathy with oculofacial apraxia. *Biochim Biophys Acta Mol Basis Dis.* 2024 Oct;1870(7):167330. doi:10.1016/j.bbadis.2024.167330. Epub 2024 Jul 1. PMID: 38960055. [Item #12.48]

Mackenzie IR, Nicholson AM, Sarkar M, Messing J, Purice MD, Pottier C, et al. TIA1 mutations in amyotrophic lateral sclerosis and frontotemporal dementia promote phase separation and alter stress granule dynamics. *Neuron* 95: 808-816, 2017. PMID: 28817800 [Item #12.78]

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

Beijer D, Marte S, Li JC, De Ridder W, Chen JZ, Tadenev ALD, et al. Dominant NARS1 mutations causing axonal Charcot-Marie-Tooth disease expand NARS1-associated diseases. *Brain Commun.* 2024 Mar 8;6(2):fcae070. doi:10.1093/braincomms/fcae070. PMID: 38495304 [Item #14.79]

Oz-Levi D, Ben-Zeev B, Ruzzo EK, Hitomi Y, Gelman A, Pelak K, et al. Mutation in TECPR2 reveals a role for

autophagy in hereditary spastic paraparesis. *Am. J. Hum. Genet.* 91: 1065-1072, 2012. PMID: 23176824 [Item #14.114]

#### **GROUP 16. MITOCHONDRIAL MYOPATHIES**

Maio N, Orbach R, Zaharieva IT, Topf A, Donkervoort S, Munot P, et al. CIAO1 loss of function causes a neuromuscular disorder with compromise of nucleocytoplasmic Fe-S enzymes. *J. Clin. Invest.* 134: e179559, 2024. PMID: 38950322 [Item #16.37]

#### **GROUP 17. OTHER NEUROMUSCULAR DISORDERS**

Tamhankar V, Tamhankar P, Chaubal R, Chaubal J, Chaubal N. Novel consensus splice site pathogenic variation in THOC2 gene leads to recurrent arthrogryposis multiplex congenita phenotype: a case report. *Cureus* 13: e19682, 2021. PMID: 34976470 [Item #17.26]

Dubucs C, Rendu J, Michel-Calemard L, Menassa R, Langeois M, Nicaise Y, et al. Muscular phenotype description of abnormal THOC2 splicing. *Neuromuscul Disord.* 33: 978-982, 2023. PMID: 37945483 [Item #17.26]