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The 2019 version of the gene table of neuromuscular disorders (nuclear genome)

Gisèle Bonne[a,*], François Rivier[b], Dalil Hamroun[c]

[a] Sorbonne Université, INSERM UMRS_974, Centre de Recherche en Myologie, Institut de Myologie, G.H. Pitié-Salpêtrière, Paris, France
[b] Neuropédiatrie & CR Maladies Neuromusculaires, CHU de Montpellier, U1046 INSERM, UMR9214 CNRS, Université de Montpellier, France
[c] CHRU de Montpellier, Direction de la Recherche et de l'Innovation, Hôpital La Colombière, Montpellier, France

General features

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

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

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

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

The two versions of the gene table[3]

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

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

* Corresponding author. Sorbonne Université, INSERM UMRS974, Centre de Recherche en Myologie, Paris, France. Fax: +33 1 42 16 57 00.
E-mail address: g.bonne@institut-myologie.org (G. Bonne).

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

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

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

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Overview of the new data in the 2019 printed version of the gene table (pages 1152 to 1183 of this issue)

There are 51 new items, marked by background shading. Altogether they comprise **26 additional genes** and **25 additional phenotypic variants** caused by a gene already listed in the 2018 version (see box). **Three previously identified loci** were solved with mutation in know genes and, finaly one locus, previously identified, but still without identifed gene was missing in the gene table (item # 10.93).

The new key references of the printed version of the table are listed in pages 1182–1183 in this issue.

Of note, two recent reports proposed revision of nomenclature for LGMD (group 1) (Straub et al. 2018) and for CMT (group 14) (Maguy et al. 2018). We decided not to implement these proposed nomenclatures in the present released printed version of the gene table of neurouscular disorders, in order to allow time for the neuromuscular community to fully validate these proposed nomenclatures.

Citation of the gene table

- Printed version: Bonne G, Rivier F, Hamroun D. The 2018 version of the gene table of neuromuscular disorders. Neuromuscul Disord. 27 (12), 1152–1183.
- Online version: GeneTable of Neuromuscular Disorders: http://www.musclegenetable.fr

Contact

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

Acknowledgements

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

New in the 2018 printed version of the gene table

26 genes added:

ALPK3 (item # 10.28)
ANXA11 (item # 12.60)
ATP1A1 (item # 14.68)
CCDC78 (item # 3.27)
CLTCL1 (item # 14.102)
COL25A1 (item # 16.8)
COX15 (item # 10.36)
DGUOK (item # 16.32)
FLVCR1 (item # 14.101)
GOSR2 (item #2.41)
HNRNPA2B1 (item # 5.27)
MIB1 (item # 10.98)
MYBPC1 (item # 16.10)
NEK1 (item # 12.61)
PIEZO2 (item # 16.16 and item # 16.17)
PRUNE1 (item # 12.77)
PSEN1 (item # 10.56)
RNASEH1 (item # 16.30)
RPH3A (item # 11.38)
RYR3 (item # 3.13)
SCO2 (item # 10.35 and item # 14.84)
SLC16A (item # 9.17)
SLC25A1 (item # 11.37)
STAC3 (item # 3.51)
UNC13 (item # 11.36)
ZFHX2 (item # 14.103)

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

ACTA1 (item # 10.95)
DES (item # 5.12)
DNAJB6 (item # 4.20)
DOK7 (item # 16.22)
FLNC (item # 10.27 and item # 10.89)
GDAP1 (item # 14.74)
LDB3 (item # 10.94)
LRSAM1 (item # 14.56)
MFN2 (item # 14.69)
MUSK (item # 16.21)
MYBPC3 (item # 10.101)
MYH7 (item # 10.96 and item # 3.35)
MYPN (item # 10.88)
NEFL (item # 14.18)
POLG (item # 16.29)
PRDM16 (item # 10.99)
RAPSN (item # 16.23)
SQSTM1 (item # 4.19)
TK2 (item # 16.31)
TNNI3 (item # 10.67)
TNNT2 (item # 10.87 and item # 10.97)
TPM1 (item # 10.100)

3 known genes for a previously identified locus

DES (item # 5.12)
LRSAM1 (item # 14.56)
MYH7 (item # 3.35)

54 new key references

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Gene table of monogenic neuromuscular disorders (nuclear genome only)
Vol. 28 No. 12, December 2018

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

Shaded background indicates newly added items.

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [M])	Key references	Other allelic disease(s) (group in this table)
GROUP 1. MUSCULAR DYSTROPHIES								
Duchenne muscular dystrophy; Becker muscular dystrophy	1.1	XR	DMD 310200 BMD 300376	Xp21.2-p21.1	***DMD*** *300377*	Dystrophin	Monaco et al (1986) Burghes et al (1987) Koenig et al (1987, 1988) Hoffman et al (1987, 1988)	allelic to CMD3B (group10)
Emery-Dreifuss muscular dystrophy, X-linked, type 1	1.2	XR	EDMD1 310300	Xq28	***EMD*** *300384*	Emerin	Hodgson et al (1986) Romeo et al (1988) Bione et al (1994, 1995) Klauck et al (1995) Nigro et al (1995)	
Emery-Dreifuss muscular dystrophy, X-linked, type 2	1.3	XR	EDMD6 300696	Xq26.3	***FHL1*** *300163*	Four and a half LIM domain 1	Gueneau et al (2009)	allelic to XPMA (group 5), SPM (group 5), RBMX1A/B (group 5)
Emery-Dreifuss muscular dystrophy, autosomal dominant	1.4	AD	EDMD2 181350	1q22	***LMNA*** *150330*	Lamin A/C	Bonne et al (1999) Worman and Bonne (2007)	allelic to EDMD3 (group 1), LGMD1B (group1), MDCL (group 2), CMD1A (group 10), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370]
Emery-Dreifuss muscular dystrophy, autosomal recessive	1.5	AR	EDMD3 616516	1q22	***LMNA*** *150330*	Lamin A/C	Raffaele di Barletta et al (2000) Worman and Bonne (2007)	allelic to EDMD2 (group 1), LGMD1B (group1), MDCL (group 2), CMD1A (group 10), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2#151660, HGPS#176670, restrictive dermopathy#275210, MADA#248370]
Nesprin-1 related muscular dystrophy	1.6	AD	EDMD4 612998	6q25.2	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Zhang et al (2007)	allelic to dilated cardiomyopathy with nesprin-1 defect (group 10), SCAR8 (group 13), AMC with nesprin-1 defect (group 16)
Nesprin-2 related muscular dystrophy	1.7	AD	EDMD5 612999	14q23.2	***SYNE2*** *608442*	Spectrin repeat containing, nuclear envelope 2 (nesprin-2)	Zhang et al (2007)	
LUMA related muscular dystrophy	1.8	AD	EDMD7 614302	3p25.1	***TMEM43*** *(=LUMA) 612048*	Transmembrane protein 43 (=LUMA)	Liang et al (2011)	allelic to ARVD5 (group 10)
LAP1B related muscular dystrophy	1.9	AR	LGMD2Y 617072	1q25.2	***TOR1AIP1*** *(=LAP1B) 614512*	Torsin A interacting protein 1 (=Lamin Associated Peptide 1B)	Kayman-Kurekci et al (2014)	

(*continued on next page*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [M])	Key references	Other allelic disease(s) (group in this table)
Facio-scapulo-humeral muscular dystrophy, type 1	1.10	AD	FSHD1 158900	4q35	***DUX4**** *606009 (*inappropriate reactivation)*	Double homeobox 4	Wijmenga et al (1990-1993) Upadhyaya et al. (1990, 1992) Wright et al (1993) van Deutekom et al (1993) Gabellini et al (2002) Van der Maarel et al (2005) Gabellini et al (2006) Petrov et al (2006) Lemmers et al (2010)	
Facio-scapulo-humeral muscular dystrophy, type 2	1.11	AD	FSHD2 158901	18p11.32	***SMCHD1**** *614982 (*causing inappropriate reactivation of DUX4* 606009)*	Structural maintenance of chromosomes flexible hinge domain containing 1	de Greef et al (2010) Sacconi et al (2012) Lemmers et al (2012) Sacconi et al (2013)	allelic to Bosma Arhinia Microphthalmia Syndrome; BAMS (#603457)
Muscular dystrophy with generalized lipodystrophy	1.12	AD		17q21.2	***CAVIN1*** *603198*	Caveolae-associated protein1, Cavin-1, (Polymerase I and transcript release factor)	Hayashi et al (2009)	
Limb girdle muscular dystrophies, dominant								
LGMD1A	1.13	AD	LGMD1A 159000	5q31	***MYOT*** *604103*	Myotilin (titin immunoglobulin domain protein)	Speer et al. (1992) Hauser et al (2000)	allelic to distal myotilinopathy (group 4), MFM (group 5), spheroid body myopathy (group 5)
LGMD1B	1.14	AD	LGMD1B 159001	1q22	***LMNA*** *150330*	Lamin A/C	van der Kooi et al (1997) Muchir et al (2000) Worman and Bonne (2007)	allelic to EDMD2 and EDMD3 (group 1), MDCL (group 2), CMD1A (group 10), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
LGMD1C	1.15	AD	LGMD1C 607801	3p25.3	***CAV3*** *601253*	Caveolin-3	Minetti et al (1998) McNally et al (1998)	allelic to MPDT (group 4), hyper CKemia (group 5), RMD2 (group 6), CMH (group 10), LQT9 (group 10)
LGMD1E (formerly LGDM1D)	1.16	AD	LGMD1E 603511	7q36.3	***DNAJB6*** *611332*	Hsp40 homologue, subfamily B, number 6	Speer et al (1999), Sarparanta e al (2012) Harms et al (2012)	allelic to distal myopathy (group 4)
LGMD1 related to *DES* (formerly LGDM1E)	1.17	AD	MFM1 601419	2q35	***DES*** *125660*	Desmin	Messina et al (1997) Greenberg et al (2012) Hedberg et al (2012)	allelic to MFM1 (group 5) and CMD1I (group 10A), ARVD7 (group 10B)
LGMD1F	1.18	AD	LGMD1F 608423	7q32.1	***TNPO3*** *610032*	Transportin 3	Palenzuela et al (2003) Melià et al (2013) Torella et al (2013)	
LGMD1G	1.19	AD	LGMD1G 609115	4q21.22	***HNRNPDL*** *607137*	Heterogeneous nuclear ribonucleoprotein D-like	Starling et al (2005) Vieira et al (2014)	
LGMD1H	1.20	AD	LGMD1H 613530	3p25.1-p23	*?*		Bisceglia et al (2010)	
Limb girdle muscular dystrophies, recessive								
LGMD2A	1.21	AR	LGMD2A 253600	15q15.1	***CAPN3*** *114240*	Calpain-3	Beckmann et al. (1991) Young et al. (1992), Richard et al (1995, 1997)	
LGMD2B	1.22	AR	LGMD2B 253601	2p13.2	***DYSF*** *603009*	Dysferlin	Bashir et al (1994) Bashir et al (1998) Liu et al (1998)	allelic to MM (group 4)
LGMD2C	1.23	AR	LGMD2C 253700	13q12.12	***SGCG*** *608896*	Gamma-sarcoglycan	Ben Othmane et al. (1992) Azibi et al. (1993) Noguchi et al (1995) McNally et al (1996) Piccolo et al (1996)	
LGMD2D	1.24	AR	LGMD2D 608099	17q21.33	***SGCA*** *600119*	Alpha-sarcoglycan	Roberds et al (1994) Piccolo et al (1995) Passos-Bueno et al (1995) Ljunggren et al (1995) Carrié et al (1997)	

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LGMD2E	1.25	AR	LGMD2E 604286	4q12	***SGCB*** *600900*	Beta-sarcoglycan	Lim et al (1995) Bönnemann et al (1995) Bönnemann et al (1996)	
LGMD2F	1.26	AR	LGMD2F 601287	5q33.3-q33.3	***SGCD*** *601411*	Delta-sarcoglycan	Passos-Bueno et al (1996) Nigro et al (1996)	allelic to CMD1L (group 10)
LGMD2G	1.27	AR	LGMD2G 601954	17q12	***TCAP*** *604488*	Titin-cap (telethonin)	Moreira et al (1997) Moreira et al (2000)	allelic to congenital muscular dystrophy with telethonin defect (group 2), CMD1N (group 10)
LGMD2H	1.28	AR	LGMD2H 254110	9q33.1	***TRIM32*** *602290*	Tripartite motif-containing 32	Weiler et al (1998) Frosk et al (2002)	allelic to sarcotubular myopathy (group 3)
LGMD2I	1.29	AR	MDDGC5 607155	19q13.32	***FKRP*** *606596*	Fukutin related protein	Driss et al (2000) Brockington et al (2001a)	allelic to MDC1C (group 2), WWS (group 2), MEB (group 2)
LGMD2J	1.30	AR	LGMD2J 608807	2q31.2	***TTN*** *188840*	Titin	Hackman et al (2003)	allelic to congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), HMERF (group 5), CMH9 (group 10), CMD1G (group 10)
LGMD2K	1.31	AR	MDDGC1 609308	9q34.13	***POMT1*** *607423*	Protein 0-mannosyl-transferase 1	Balci et al (2005) D'Amico et al (2006)	allelic to WWS (group 2)
LGMD2L	1.32	AR	LGMD2L 611307	11p14.3	***ANO5*** *(TMEM16E)* *608662*	Anoctamin 5	Jarry et al (2007), Bolduc et al (2008, 2010), Hicks et al (2011)	allelic to early onset calf distal myopathy (group 4)
LGMD2M	1.33	AR	MDDGC4 611588	9q31.2	***FKTN*** *607440*	Fukutin	Murakami et al (2006) Godfrey et al (2006)	allelic to FCMD (group 2), WWS (group 2), CMD1X (group 10)
LGMD2N	1.34	AR	MDDGC2 613158	14q24.3	***POMT2*** *607439*	Protein O-mannosyl transferase 2	Biancheri et al (2007)	allelic to WWS (group 2) and to MEB (group 2)
LGMD2O	1.35	AR	MDDGC3 613157	1p34.1	***POMGNT1*** *606822*	Protein O-linked mannose beta1,2-N-acetylglucos-aminyl-transferase 1	Godfrey et al (2007); Clement et al (2008) Raducu et al (2012)	allelic to WWS (group 2) and to MEB (group 2)
LGMD2P	1.36	AR	MDDGC9 613818	3p21.31	***DAG1*** *128239*	Dystrophin-associated glycoprotein 1 (alpha-dystroglycan)	Hara et al (2011)	allelic to MDDGA9 (group 2)
LGMD2Q	1.37	AR	LGMD2Q 613723	8q24.3	***PLEC*** *601282*	Plectin	Gundesli et al (2010)	allellic to LGMD with ophthalmoplegia (group 1), MDEBS (group 5), and Myasthenic syndrome with plectin defect (group 11)
LGMD2R	1.38	AR	LGMD2R 615325	2q35	***DES*** *125660*	Desmin	Cetin et al (2013)	allelic to other desminopathies (groups 1, 5 and 10)
LGMD2S	1.39	AR	LGMD2S 615356	4q35.1	***TRAPPC11*** *614138*	Trafficking protein particle complex 11	Bögershausen et al (2013)	allelic to CMD related to TRAPPC11 (group 2)
LGMD2T	1.40	AR	MDDGC14 615352	3p21.31	***GMPPB*** *615320*	GDP-mannose py-rophosphorylase B	Carss et al (2013), Cabrera-Serrano et al (2015)	allelic to MDDGA14, MDDGB14 (group 2) and congenital myyasthenic syndrome (group 11)
LGMD2U	1.41	AR	MDDGC7 616052	7p21.2-p21.1	***ISPD*** *614631*	Isoprenoid synthase domain containing protein	Tasca et al (2013)	allelic to WWS/ MDDGA7 (group 2)
LGMD2V	1.42	AR		17q25.3	***GAA*** *606800*	Glucosidase alpha, acid	Preisler et al (2013)	allelic to Pompe's disease (groups 9 and 10)
LGMD2W	1.43	AR	LGMD2W 616827	2q14.3	***LIMS2*** *(= PINCH2)* *607908*	LIM and senescent cell antigen-like domains 2	Chardon et al (2015)	
LGMD2X	1.44	AR	LGMD2X 616812	6q21	***BVES*** *(= POPDC1)* *604577*	Blood vessel epicardial substance	Schindler et al (2016)	
LGMD2Y	1.45	AR	LGMD2Y 617072	1q25.2	***TOR1AIP1*** *614512*	Torsin 1A interacting protein 1 (= lamin associated protein 1)	Kayman-Kurekci et al (2014); Sewry et al (2014)	
LGMD2Z	1.46	AR	LGMD2Z 617232	3q13.33	***POGLUT1*** *615618*	Protein O-Glucosyl-transferase 1	Servian-Morilla et al (2016)	

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Muscle dystrophy with glycosylation defect, type Io	1.47	AR	CDG1O 612937	1q22	***DPM3*** *605951*	Dolichyl-phosphate mannosyltrans-ferase polypeptide 3	Lefeber et al (2009)	
Scapuloperoneal muscular dystrophy and dropped head syndrome	1.48	AR	600416	9p13.3	***VCP*** *601023*	Valosin-containing protein	Liewluck et al (2014)	allelic to IBMPFD (groups 4 and 5), ALS14 (group 12) and CMT2Y (group 14)
LGMD with ophthalmoplegia	1.49	AR		8q24.3	***PLEC*** *601282*	Plectin	Fattahi et al (2015)	allelic to LGMD2Q (group 1), MDEBS (group 5), myasthenic syndrome with plectin defect (group 11)
GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES								
Congenital muscular dystrophy with merosin deficiency	2.1	AR	MDC1A 607855	6q22.33	***LAMA2*** *156225*	Laminin 2, Heavy chain (laminin alpha2 chain of merosin)	Tomé et al (1994) Hillaire et al (1994) Helbling-Leclerc et al (1995) Allamand et al (1997)	
Bethlem myopathy	2.2	AD	BTHLM1 158810	21q22.3	***COL6A1*** *120220*	Collagen type VI subunit alpha 1	Jöbsis et al (1996)	allelic to UCMD (group 2)
Bethlem myopathy	2.3	AD	BTHLM1 158810	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Jöbsis et al (1996)	allelic to UCMD (group 2), and myosclerosis (group 2)
Bethlem myopathy	2.4	AD	BTHLM1 158810	2q37.3	***COL6A3*** *120250*	Collagen type VI subunit alpha 3	Speer et al (1996) Bertini et al (1998) Pan et al (1998)	allelic to UCMD (group 2)
Bethlem myopathy	2.5	AR	BTHLM1 158810	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Gualandi et al (2009)	allelic to UCMD (group 2)
Ullrich congenital muscular dystrophy	2.6	AR	UCMD 254090	21q22.3	***COL6A1*** *120220*	Collagen, type VI, subunit alpha 1	Pan et al (2003) Giusti et al (2005)	allelic to Bethlem myopathy (group 2)
Ullrich congenital muscular dystrophy	2.7	AR	UCMD 254090	21q22.3	***COL6A2*** *120240*	Collagen, type VI, subunit alpha 2	Vanegas et al (2001) Higuchi et al (2001)	allelic to Bethlem myopathy (group 2) and myosclerosis (group 2)
Ullrich congenital muscular dystrophy	2.8	AR	UCMD 254090	2q37.3	***COL6A3*** *120250*	Collagen type VI subunit alpha 3	Demir et al (2002)	allelic toallelic to Bethlem myopathy (group 2) and myosclerosis (group 2)
Ullrich congenital muscular dystrophy 2	2.9	AR	UCMD2 616470	6q13-q14	***COL12A1*** *120320*	Collagen type XII alpha 1 chain	Zou et al (2014)	
Bethlem myopathy 2	2.10	AD	BTHLM2 616471	6q13-q14	***COL12A1*** *120320*	Collagen type XII alpha 1 chain	Zou et al (2014)	
COL12A1-related congenital muscular dystrophy	2.11	AD		6q13-q14	***COL12A1*** *120320*	Collagen type XII alpha 1 chain	Punetha et al (2016)	
Myosclerosis	2.12	AR	255600	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha 2	Merlini et al (2008)	allelic to UCMD (group 2) and to Bethlem myopathy (group 2)
Rigid spine syndrome related to *SEPN1*	2.13	AR	RSMD1 602771	1p36.11	***SELENON*** *(formerly SEPN1)* *606210*	Selenoprotein N	Moghadaszadeh et al (1998, 2001) Ferreiro et al (2002, 2004)	allelic to CFTD (group 3), multiminicore disease (group 3), and desminrelated myopathy with Mallory bodies (group 5)
Congenital muscular dystrophy related to integrin	2.14	AR	613204	12q13.2	***ITGA7*** *600536*	Integrin alpha7	Hayashi et al (1998)	
Congenital muscular dystrophy related to dynamin 2	2.15	AD		19p13.2	***DNM2*** *602378*	Dynamin 2	Susman et al (2008)	allelic to CNM1 (group 3) and CMTIB (group 14)
Congenital musuclar dystrophy related to telethonin	2.16	AR		17q12	***TCAP*** *604488*	Titin-cap (telethonin)	Ferreiro et al (2011)	allelic to LGMD2G (group 1) and to CMD1N (group 10A)
Congenital muscular dystrophy related to *LMNA*	2.17	AD	MDCL 613205	1q22	***LMNA*** *150330*	Lamin A/C	Quijano-Roy et al (2008)	allelic to EDMD2 and EDMD3 (group 1), MDCL (group 2), CMD1A (group 10), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
CMD-Dystroglycanopathies								
Fukuyama congenital muscular dystrophy	2.18	AR	MDDGA4 253800 MDDGB4 613152	9q31.2	***FKTN*** *607440*	Fukutin	Toda et al. (1993) Kobayashi et al (1998)	allelic to WWS (group 2)
Walker-Warburg syndrome (WWS)	2.19	AR	MDDGA4 253800 MDDGB4 613152	9q31.2	***FKTN*** *607440*	Fukutin	Beltran-Valero de Bernabe (2003) Mercuri et al (2009)	allelic to LGMD2M (group 1) and Fukuyama (group 2) and CMD1X (group 10)
Walker-Warburg syndrome (WWS)	2.20	AR	MDDGA1 236670 MDDGB1 607423	9q34.13	***POMT1*** *607423*	Protein-O-mannosyl transferase 1	Beltran-Valero De Bernabe et al (2002) van Reeuwijk et al (2006) Mercuri et al (2009)	allelic to LGMD2K (group 1)

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Walker-Warburg syndrome (WWS)	2.21	AR	MDDGA2 613150 MDDGB2 613156	14q24.3	***POMT2*** *607439*	Protein O-mannosyl transferase 2	van Reeuwijk et al (2005) Mercuri et al (2009)	allelic to LGMD2N (group 1) and MEB (group 2)
Walker-Warburg syndrome (WWS)	2.22	AR	MDDGA5 613153 MDDGB5 606612	19q13.32	***FKRP*** *606596*	Fukutin related protein	Beltran-Valero De Bernabe et al (2004)	allelic to LGMD2I (group 1), MDC1C (group 2), MEB (group 2)
Walker-Warburg syndrome (WWS)	2.23	AR	MDDGA3 253280 MDDGB3 613151	1p34.1	***POMGNT1*** *606822*	Protein O-mannose beta1,2-N-acetylglucos-aminyl transferase	Taniguchi et al (2003) Mercuri et al (2009)	allelic to MEB (group 2)
Walker-Warburg syndrome (WWS)	2.24	AR	MDDGA7 614643	7p21.2-p21.1	***ISPD*** *614631*	Isoprenoid synthase domain containing	Roscioli et al (2012) Willer et al (2012)	
Walker-Warburg syndrome (WWS)	2.25	AR	MDDGA8 614830	3p22.1	***POMGNT2*** *(formerly GTDC2) 614828*	Protein O-mannose beta-1,4-N-acetylglucos-aminyl transferase 2	Manzini et al (2012)	
Walker-Warburg syndrome (WWS)	2.26	AR	MDDGA13 615287	11q13.2	***B4GAT1*** *(formerly B3GNT1) 605517*	Beta-1,4-glucuronyl transferase 1 (Beta-1,3-N-acetylglucos-aminyltransferase 1)	Buysse et al (2013) Shaheen et al (2013)	
Muscle-eye-brain disease (MEB)	2.27	AR	MDDGA3 253280	1p34.1	***POMGNT1*** *606822*	Protein O-mannose beta1,2-N-acetylglucos-aminyl transferase	Yoshida et al (2001) Taniguchi et al (2003)	allelic to WWS (group 2)
Muscle-eye-brain disease (MEB)	2.28	AR	MDDGA5 613153 MDDGB5 606612	19q13.32	***FKRP*** *606596*	Fukutin related protein	Beltran-Valero De Bernabe et al (2004)	allelic to LGMD2I (group 1), MDC1C (group 2), WWS (group 2)
Muscle-eye-brain disease (MEB)	2.29	AR	MDDGA2 613150 MDDGB2 613156	14q24.3	***POMT2*** *607439*	Protein O-mannosyl transferase 2	Mercuri et al (2006)	allelic to WWS (group 2)
Muscle-eye-brain disease (MEB)	2.30	AR	MDDGC14 615350	3p21.31	***GMPPB*** *615320*	GDP-mannose py-rophosphorylase B	de Carss et al (2013)	allelic to LGMD2T (group 1) and MDDGB14 (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.31	AR	MDDGA5 613153 MDDGB5 606612	19q13.32	***FKRP*** *606596*	Fukutin related protein	Brockington et al (2001b) Topaloglu et al (2003) Mercuri et al (2009)	allelic to LGMD2I (group 1), WWS, MEB (group 2) and congenital myasthenic syndrome (group 11)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.32	AR	MDDGA6 613154 MDDGB6 608840	22q12.3	***LARGE*** *603590*	Acetylglucos-aminyl-transferase-like protein (like-glycosyl transferase)	Longman et al (2003) Mercuri et al (2009)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.33	AR	CDG1E 608799	20q13.13	***DPM1*** *603503*	Dolichyl-phosphate mannosyltrans-ferase 1, catalytic subunit	Yang et al (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and severe epilepsy	2.34	AR	CDG1U 615042	9q34.11	***DPM2*** *603564*	Dolichyl-phosphate mannosyltrans-ferase polypeptide 2, regulatory subunit	Barone et al (2012)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.35	AR	MDDGA9 616538	3p21.31	***DAG1*** *128239*	Dystrophin-Associated Glycoprotein 1	Geis et al (2013)	allelic to LGMD2P (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A10	2.36	AR	MDGGA10 615041	12q14.2	***RXYLT1*** *(formerly TMEM5) 605862*	Ribitol xylosyltransferase 1 (transmembrane protein 5)	Vuillaumier-Barrot et al (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan WWWS/MEB like	2.37	AR	MDDGA11 615181	1q42.3	***B3GALNT2*** *610194*	Beta-1,3-N-acetylgalactos-aminyltransferase 2	Stevens et al (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.38	AR	MDDGA12 615249	8p11.21	***POMK*** *615247*	Protein-O-mannose kinase	Jae et al (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and mental retardation	2.39	AR	MDDGB14 615351	3p21.31	***GMPPB*** *615320*	GDP-mannose py-rophosphorylase B	Carss et al (2013)	allelic to LGMD2T (group 1), MDDGC14 (group 2)

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Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by *TRAPPC11*mutations	2.40	AR		4q35.1	***TRAPPC11*** *614138*	Trafficking protein particle complex 11	Liang et al (2015) Larson et al (2018)	allelic to LGMD2S (group 1)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and epilepsy	2.41	AR		17q21.32	***GOSR2*** *604027*	Golgi SNAP receptor complex member 2	Larson et al (2018)	
Other congenital muscular dystrophies								
Congenital muscle dystrophy with joint hyperlaxity	2.42	AR		3p23-21	*?*		Tetreault et al (2006)	
Congenital muscle dystrophy with mitochondrial structural abnormalities (megaconial type)	2.43	AR	MDCMC 602541	22q13.33	***CHKB*** *612395*	Choline kinase beta	Mitsuhashi et al (2011)	
Congenital muscular dystrophy	2.44	AR	MDC1B 604801	1q42	*?*		Brockington et al (2000)	
Congenital muscular dystrophy with rigid spine related to *ACTA1*	2.45	AR	Possibly identical to MDC1B 604801	1q42.13	***ACTA1*** *102610*	Alpha actin, skeletal muscle	O'Grady et al. (2014)	allelic to NEM3, CFTD, cap myopathy related to ACTA1 (group 3)
GOLGA2-related congenital muscle dystrophy with brain involvement	2.46	AR		9q34.11	***GOLGA2*** *602580*	Golgin A2	Shamseldin et al (2016)	
Muscular dystrophy, congenital Davignon-Chauveau type	2.47	AR	MDCDC 617066	15q22.31	***TRIP4*** *604501*	Thyroid hormone receptor interactor 4	Davignon et al (2016)	allelic to SMABF1 (group 12)
Muscular dystrophy, congenital, with cataracts and intellectual disability	2.48	AR	MDCCAID 617404	17p13.3	***INPP5K*** *607875*	Inositol Polyphosphate-5-Phosphatase K	Osborn et al (2017) Wiessner et al (2017)	
GROUP 3. CONGENITAL MYOPATHIES								
Nemaline myopathies								
NEM1	3.1	AD	NEM1 609284	1q21.3	***TPM3*** *191030*	Tropomyosin 3	Laing et al. (1992) Laing et al. (1995b) Tan et al (1999) Wattanasirichaigoon et al (2002)	allelic to CFTD (group 3)
NEM2	3.2	AR	NEM2 256030	2q22.3	***NEB*** *161650*	Nebulin	Wallgren-Pettersson et al (1995, 2002) Pelin et al (1999) Lehtokari et al (2006)	allelic to distal myopathy (group 4)
NEM3	3.3	AD	NEM3 161800	1q42.13	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Nowak et al (1999)	allelic to CFTD (group 3)
NEM4	3.4	AD	NEM4 609285	9p13.3	***TPM2*** *190990*	Tropomyosin 2 (beta)	Donner et al (2002) Monnier et al (2009)	allelic to DA2B and DA1 (group 16)
NEM5	3.5	AR	NEM5 605355	19q13.42	***TNNT1*** *191041*	Troponin T type 1 (skeletal, slow)	Johnston et al (2000	
NEM6	3.6	AD	NEM6 609273	15q22.31	***KBTBD13*** *613727*	Kelch repeat and BTB/POZ domain containing protein 13	Gommans et al (2003) Samburghin et al (2010)	
NEM7	3.7	AR	NEM7 610687	14q13.1	***CFL2*** *601443*	Cofilin 2 (muscle)	Agrawal et al (2007)	
NEM8	3.8	AR	NEM8 615348	3p22.1	***KLHL40*** *615340*	Kelch-like family member 40	Ravenscroft et al (2013)	
NEM9	3.9	AR	NEM9 615731	2q31.1	***KLHL41*** *607701*	Kelch-like family member 41	Gupta et al (2013)	
NEM10	3.10	AR	NEM 10 616165	3p14.1	***LMOD3*** *616112*	Leiomodin 3	Yuen et al (2014)	
NEM11	3.11	AR	NEM11 617336	10q21.3	***MYPN*** *608517*	Myopalladin	Miyatake et al (2016) Lornage et al (2017)	allelic to CMD1KK, CMD22 and RCM 4 (group 10A)
Nemaline myopathy with cardiomyopathy	3.12	AR		22q12.1	***MYO18B*** *607295*	Myosin XVIIIB	Malfatti et al (2015)	allelic to KFS4 (#616549)
Myopathy with nemaline bodies	3.13	AR		15q13-q14	***RYR3*** *180903*	Ryanodine receptor 3	Nilipour et al (2018)	
Other congenital myopathies								
Myopathy, congenital, with fiber-type disproportion	3.14	AD, AR	CFTD 255310	1q42.13	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Clarke et al (2003) Laing et al (2004)	allelic to NEM3 (group 3)
Myopathy, congenital, with fiber-type disproportion	3.15	AD, AR	CFTD 255310	1p36.11	***SELENON*** *(formerly SEPN1) 606210*	Selenoprotein N	Clarke et al (2006)	allelic to RSMD1 (group 2), CFTD, multminicore disease (group 3), desmin-related myopathy with Mallory bodies (group 5)
Myopathy, congenital, with fiber-type disproportion	3.16	AD, AR	CFTD 255310	1q21.3	***TPM3*** *191030*	Tropomysosin 3	Clarke et al (2008)	allelic to NEM1 (group 3)

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Myopathy, congenital, with fiber-type disproportion	3.17	AR	CFTD 255310	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Clarke et al (2010)	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Myopathy congenital, with fiber-type disproportion	3.18	AD	CFTD 255310	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Ortolano et al (2011)	allelic to MSMA (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10)
Myopathy congenital, with fiber-type disproportion	3.19	AR	CNM6 617760	2q31.1	***ZAK*** *609479*	Leucine zipper-and sterile alpha motif-containing kinase	Vasli et al (2017)	
Myotubular myopathy	3.20	XR	CNMX 310400	Xq28	***MTM1*** *300415*	Myotubularin 1	Thomas et al (1987) Laporte (1996, 1997, 2000)	
Centronuclear myopathy related to *DNM2*	3.21	AD	CNM1 160150	19p13.2	***DNM2*** *602378*	Dynamin 2	Bitoun et al (2005)	allelic to CMD related to dynamin 2 (group 2) and CMTDIB (group 14)
Centronuclear myopathy related to *BIN1*, recessive	3.22	AR	CNM2 255200	2q14.3	***BIN1*** *601248*	Bridging integrator 1 (Amphiphysin)	Nicot et al (2007) Böhm et al (2014)	
Centronuclear myopathy related to *BIN1,* dominant	3.23	AD		2q14.3	***BIN1*** *601248*	Bridging integrator 1 (Amphiphysin)	Böhm et al (2014)	
Centronuclear myopathy related to *RYR1*	3.24	AR	255200	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Wilmshurst et al (2010)	allelic to CFTD related to RYR1, CCD, minicore myopathy with external ophthalmoplegia (group 3), late onset axial myopathy (group 5), MHS1 (group 8)
Centronuclear myopathy related to *TTN*	3.25	AR		2q31.2	***TTN*** *188840*	Titin	Ceyhan-Birsoy et al (2013)	allelic to LGMD2J (group 1), SALMY (group 3), TMD (group 4) HMERF (group 5), CMH9 (group 10), CMD1G (group 10)
Centronuclear myopathy with dilated cardiomyopathy	3.26	AR	CNM5 615959	2q35	***SPEG*** *615950*	SPEG complex locus	Agrawal et al (2014)	
Congenital myopathy with cores and central nuclei	3.27	AD	CNM4 614807	16p13.3	***CCDC78*** *614666*	Coiled-coil domain-containing protein 78	Majczenko et al (2012)	
Central core disease, dominant	3.28	AD	CCD 117000	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Kausch et al (1991) Zhang et al (1993) Quane et al. (1993) Robinson et al (2002)	allelic to CFTD & centronuclear myopathy related to RYR1, minicore myopathy with external ophthalmoplegia (group 3), late onset axial myopathy (group 5), MHS1 (group 8)
Central core disease, recessive (transient multiminicore myopathy)	3.29	AR	CCD 117000	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Ferreiro et al (2002a) Jungbluth et al (2002)	allelic to CFTD & centronuclear myopathy related to RYR1, minicore myopathy with external ophthalmoplegia (group 3), late onset axial myopathy (group 5), MHS1 (group 8)
Minicore myopathy with external ophthalmoplegia	3.30	AR	255320	19q13.2	***RYR1*** *180901*	Ryanodine receptor	Monnier et al (2003) Jungbluth et al (2005)	allelic to CFTD & centronuclear myopathy related to RYR1, minicore myopathy with external ophthalmoplegia (group 3), late onset axial myopathy (group 5), MHS1 (group 8)
Multiminicore disease, classical form	3.31	AR	255320	1p36.11	***SELENON*** *(formerly SEPN1) 606210*	Selenoprotein N	Ferreiro et al (2002) Ferreiro et al (2004)	allelic to RSMD1 (group 2), desmin related myopathy with Mallory bodies (group 5)
Early onset myopathy, areflexia, respiratory distress and dysphagia	3.32	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	Multiple EGF-like-domains 10	Logan et al (2011)	
Recessive congenital myopathy with minicores	3.33	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	Multiple EGF-like-domains 10	Boyden et al (2012)	
Myopathy, myosin storage, autosomal dominant	3.34	AD	MSMA 608358	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Tajsharghi et al (2003) Bohlega et al (2004) Laing et al (2005)	allelic to CFTD (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10)
Myopathy, myosin storage, autosomal recessive	3.35	AR	MSMB 255160	14q11.2	***MYH7*** *160780*	Myosin, heavy chain 7, cardiac muscle, beta	Onengut et al (2004) Tajsharghi et al (2007) Yuceyar et al (2015)	allelic to CFTD (group 3), MPD1 (group 4), CMH1, CMD1S and LVNC5 (group 10)
Myopathy, proximal, and ophthalmoplegia (inclusion body myopathy 3)	3.36	AD, AR	MYPOP 605637	17p13.1	***MYH2*** *160740*	Myosin, heavy chain 2, skeletal muscle, adult	Martinsson et al (1999, 2000) Tajsharghi et al (2010)	
Isolated inclusion body myopathy	3.37	AD	IBMPFD3 615424	12q13.13	***HNRNPA1*** *164017*	Heterogeneous nuclear ribonucleoprotein A1	Izumi et al (2015)	allelic to ALS20 (group 12)

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Cap myopathy	3.38	AD	CAPM2 609285	9p13.3	***TPM2*** *190990*	Tropomyosin 2, beta	Tajsharghi et al (2007) Lehtokari et al (2007)	allelic to NEM4 (group 3), DA1 and DA2B (group 16)
Cap myopathy	3.39	AD	CAPM1 609284	1q21.3	***TPM3*** *191030*	Tropomysosin 3	De Paula et al (2009) Ohlsson et al (2009)	allelic to NEM1 (group 3)
Cap myopathy	3.40	AD		1q42.13	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Hung et al (2010)	allelic to NEM3 (group 3)
Congenital neuromuscular disease with uniform type 1 fiber	3.41	AR, AD	CCD 117000	19q13.2	*RYR1* *180901*	Ryanodine receptor I	Sato et al (2007)	allelic to CFTD & centronuclear myopathy related to RYR1, CCD, minicore myopathy with external ophthalmoplegia (group 3), late onset axial myopathy (group 5), MHS1 (group 8)
Salih myopathy, (Congenital myopathy with fatal cardiomyopathy)	3.42	AR	SALMY 611705	2q31.2	*TTN* *188840*	Titin	Carmignac et al (2007)	Allelic to LGMD2J (group 1), CNM related to TTN (groupe 3), TMD (group 4), HMERF (group 5), CMH9 (group 10), CMD1G (group 10)
Congenital skeletal myopathy and fatal cardiomyopathy	3.43	AR		11p11.2	***MYBPC3*** *600958*	Cardiac myosin binding protein-C	Tajsharghi et al (2010)	allelic to CMH4, CMD1MM and LVNC10 (group 10)
Congenital lethal myopathy	3.44	AR	MYPCN 612540	12q12	***CNTN1*** *600016*	Contactin-1	Compton et al (2008)	
Sarcotubular myopathy	3.45	AR		9q33.1	***TRIM32*** *602290*	Tripartite motif containing 32	Schoser et al (2005)	allelic to LGMD2H (group 1)
Congenital myopathy related to *PTPLA*	3.46	AR		10p12.33	***HACD1*** *(=PTPLA)* *610467*	3-Hydroxyacyl-CoA dehydratase (Protein tyrosine phosphatase-like)	Muhammad et al (2013)	allelic to ARVD6 (group 10)
Congenital myopathy with ophthalmoplegia related to *CACNA1S*	3.47	AR		1q32.1	***CACNA1S*** *114208*	Calcium channel,voltage-dependent, L type, alpha 1S subunit	Hunter et al. (2015)	Allelic to hypoKPP1 (group 7) MHS5 (group 8)
Congenital myopathy with neuropathy and deafness	3.48	AR	NEDHND 617519	19q13.2	***SPTBN4*** *606214*	Spectrin, beta, nonerythrocytic, 4	Knierim et al (2017)	
Myopathy, congenital, With excess of muscle spindles	3.49	AD	CMEMS 218040	11p15.5	***HRAS*** *190020*	V-Ha-RAS Harvey rat sarcoma viral oncogene homolog	Quélin et al (2017)	
Myopathy, Congenital nonprogressive, with Moebius sequence and Robin sequence	3.50	AR	CFZS 254940	9q34.2	***MYMK*** *615345*	Myomaker	Di Gioia et al (2017)	
Myopathy, congenital, Baily-Bloch (Native American myopathy)	3.51	AR	MYPBB 255995	12q13.3	***STAC3*** *615521*	SH3 and cysteine-rich domains 3	Horstick et al (2013)	
GROUP 4. DISTAL MYOPATHIES								
Distal recessive myopathy (Miyoshi)	4.1	AR	MMD1 254130	2p13.2	***DYSF*** *603009*	Dysferlin	Bejaoui et al . (1995) Bashir et al (1998) Liu et al (1998)	allelic to LGMD2B (group 1)
Tibial muscular dystrophy (Udd)	4.2	AD	TMD 600334	2q31.2	***TTN*** *188840*	Titin	Haravuori et al (1998, 2001) Hackman et al (2002)	allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), HMERF (group 5), CMH9 (group 10), CMD1G (group 10)
Distal myopathy with rimmed vacuoles (Nonaka) and Hereditary inclusion body myopathy	4.3	AR	NM 605820	9p13.3	***GNE*** *603824*	Glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannos-amine kinase	Mitrani-Rosenbaum et al (1996) Ikeuchi et al (1997) Eisenberg et al (2001)	
Distal myopathy (Laing)	4.4	AD	MPD1 160500	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Laing et al. (1995) Mastaglia et al (2000) Meredith et al (2004)	allelic to CFTD and myosin storage myopathy (group 3), CMH1 and CMD1S (group 10)
Vocal cord and pharyngeal distal myopathy	4.5	AD	ALS21 606070	5q31.2	***MATR3*** *164015*	Matrin 3	Feit et al (1998) Senderek et al (2009)	Allelic to ALS21 (group 12)
Adult onset distal myopathy	4.6	AD	MPD3 610099	8p22-q11	*?*		Haravuori et al (2004)	
Welander distal myopathy	4.7	AD	WDM 604454	2p13.3	***TIA1*** *603518*	Cytotoxic granule-associated RNA binding protein	Ahlberg et al (1999) Hackman et al (2013) Klar et el (2013)	
Distal myopathy with pes cavus and areflexia (Vacuolar neuromyopathy)	4.8	AD	601846	19p13.3	*?*		Servidei et al (1999)	
Distal myopathy with myotilin defect	4.9	AD		5q31.2	***MYOT*** *604103*	Myotilin	Penisson-Besnier et al (1998, 2006)	Allelic to LGMD1A (group 1), MFM (group 5), spheroid body myopathy (group 5)
Distal myopathy with nebulin defect	4.10	AR		2q23.3	***NEB*** *161650*	Nebulin	Wallgren-Pettersson et al (2007)	allelic to NEM2 (group 3)

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Distal myopathy, Tateyama type	4.11	AD	MPDT 614321	3p25.3	***CAV3*** *601253*	Caveolin-3	Tateyama et al (2002) Fulizio et al (2005)	allelic to LGMD1C (group 1), hyper CKemia (group 5), RMD2 (group 6), CMH and LQT9 (group 10)
Late onset distal myopathy (Markesbery-Griggs)	4.12	AD		10q23.2	***LDB3*** *605906*	LIM domain binding-3	Griggs et al (2007)	allelic to MFM4 (group 5), CMD1C (group 10) and CMH24 (group 10)
Early onset calf distal myopathy	4.13	AR	MMD3 613319	11p14.3	***AN05*** *608662*	Anoctamin 5	Bolduc et al. (2010)	allelic to LGMD2L (group 1)
Dynamin 2 related distal myopathy	4.14	AD	CNM1 160150	19p13.2	***DNM2*** *602378*	Dynamin 2	Fischer etal (2006)	allelic to CNM (group 3) and CMTDIB (group 14)
Early onset distal myopathy with *KLHL9* defect	4.15	AD		9p21.3	***KLHL9*** *611201*	Kelch-like 9	Cirak et al (2010)	
Filamin C related distal myopathy	4.16	AD	MPD4 614065	7q32.1	***FLNC*** *102565*	Filamin C (gamma)	Duff et al (2011)	allelic to MFM5 (group 5), CMH26 and RCM4 (group 10)
Distal myopathy with VCP defect	4.17	AD	IBMPFD1 167320	9p13.3	***VCP*** *601023*	Valosin-containing protein	Palmio et al (2011)	allelic to scapuloperoneal myoathy (group 1), IBMBFD1 (group 5), ALS14 (group 12) and CMT2Y (group 14)
Adolescent onset distal myopathy	4.18	AR	MPD5 617030	14q32.33	***ADSSL1*** *612498*	Adenylosucci-nate synthase-like	Park et al (2016)	
Myopathy, distal, with rimmed vacuoles	4.19	AD	DMRV 617158	5q35.3	***SQSTM1*** *601530*	Sequestosome 1	Bucelli et al (2015)	allelic to FTDALS3 (group 12)
Myopathy, distal, with rimmed vacuoles	4.20	AD		7q36.3	***DNAJB6*** *611332*	Hsp40 homologue, subfamily B, number 6	Ruggieri et al (2015)	allelic to LGMD1E (group 1)
GROUP 5. OTHER MYOPATHIES								
Myofibrillar myopathies								
Myofibrillar myopathy, desmin related	5.1	AD	MFM1 601419	2q35	***DES*** *125660*	Desmin	Goldfarb et al (1998) Munoz-Marmol et al (1998)	allelic to LGMD1E and LGM2R (group1), MFM1 with ARVC (group 5), CMD1I and ARVD7 (group 10)
Myofibrillar myopathy, alpha-B crystallin related	5.2	AD	MFM2 608810	11q23.1	***CRYAB*** *123590*	Alpha-B crystallin	Vicart et al (1998) Selcen et al (2003)	allelic to CMD1II (group 10)
Myofibrillar myopathy, myotilin related	5.3	AD	MFM3 609200	5q31.2	***MYOT*** *604103*	Myotilin	Selcen and Engel (2004)	allelic to LGMD1A (group 1), spheroid body myopathy (group 5)
Spheroid body myopathy	5.4	AD	182920	5q31.2	***MYOT*** *604103*	Myotilin	Foroud et al (2005)	allelic to LGMD1A (group 1), MFM3 (group 5)
Myofibrillar myopathy, ZASP related	5.5	AD	MFM4 609452	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Selcen and Engel (2005)	allelic to Markesbery-Griggs (group 4), CMD1C and CMH24 (group 10)
Myofibrillar myopathy, filamin-C related	5.6	AD	MFM5 609524	7q32.1	***FLNC*** *102565*	Filamin C (gamma)	Vorgerd et al (2005)	allelic to MPD4 (group 4), CMH26 and RCM5 (group 10)
Myofibrillar myopathy, BAG3 related	5.7	AD	MFM6 612954	10q26.11	***BAG3*** *603883*	BCL2-associated athanogene 3	Selcen et al (2009)	allelic to CMD1HH (group 10) and CMT (group 14)
Myofibrillar myopathy, KY related	5.8	AR	MFM7 617114	3q22.2	***KY*** *605739*	Kyphoscoliosis peptidase	Hedberg-Oldfors et al (2016) Straussberg et al (2016)	
Myofibrillar myopathy, PYROXD1 related	5.9	AR	MFM8 617258	12p12.1	***PYROXD1*** *617220*	Pyridine nucleotide-disulphide oxidoreductase domain 1	O'Grady et al (2016)	
Desmin-related myopathy with Mallory bodies	5.10	AD	RSMD1 602771	1p36.11	***SELENON*** *(formerly SEPN1) 606210*	Selenoprotein N	Ferreiro et al (2004)	allelic to RSMD1 (group 2), CFTD (group 3) multiminicore disease (group 3)
Cardiac and skeletal aggregate myopathy	5.11	Digenic		1p36.11 2p.23.3	***TRIM63*** *06131* + ***TRIM54*** *606474*	Tripartite motif containing 63 (MURF1) + Ring finger protein 30 (MURF3)	Olivé et al (2015)	
Myofibrillar myopathy with arrhythmogenic right ventricular cardiomyopathy	5.12	AD	MFM1 601419	10q22	***DES*** *125660*	Desmin	Melberg et al (1999) Kuhl et al (2008) Hedberg et al (2012)	allelic to LGMD1 and LGM2R (group 1), MFM1 (group 5), CMD1I and ARVD7 (group 10)
Miscellaneous								
Danon disease	5.13	XD	GSD2B 300257	Xq24	***LAMP2*** *309060*	Lysosomal-associated membrane protein 2	Nishino et al (2000) Musumeci et al (2005)	

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [M])	Key references	Other allelic disease(s) (group in this table)
Myopathy with excessive autophagy	5.14	XR	MEAX 310440	Xq28	***VMA21*** *300913*	S. Cerevisiae homolog of VMA21	Saviranta et al (1988) Villard et al (2000) Minassian et al (2002) Munteanu et al (2008) Ramachandran et al (2013) Crockett et al (2014)	
Autophagic vacuolar myopathy	5.15	AR	CLN3 204200	16p12.1	***CLN3*** *607042*	Ceroid-lipofuscinosis, neuronal 3 (= Battenin)	Cortese et al (2014)	
Oculopharyngeal muscular dystrophy	5.16	AD	OPMD 164300	14q11.2	***PABPN1*** *602279*	Polyadenylate-binding protein, nuclear 1	Brais et al (1995, 1998) Robinson et al (2005)	
Hereditary myopathy with early respiratory failure (Edström myopathy)	5.17	AD	HMERF 603689	2q31.2	***TTN*** *188840*	Titin	Nicolao et al (1999) Lange et al (2005)	allelic to LGMD2J (group 1), CMN and SALMY (group 3), TMD (group 4), CMH9 (group 10), CMD1G (group 10)
Epidermolysis bullosa simplex associated with muscular dystrophy	5.18	AR	EBSMD 226670	8q24.3	***PLEC*** *601282*	Plectin	Gache et al (1996) Smith et al (1996) Wuyts et al (1996)	allelic to LGMD2Q (group 1), myasthenic syndrome with plectin defect (group 11)
Muscle hypertrophy	5.19	AR	MSLHP 614160	2q32.2	***MSTN 601788***	Myostatin	Schuelke et al (2004)	
Fibrodysplasia ossificans progressiva	5.20	AD	FOP 135100	2q24.1	***ACVR1*** *102576*	Activin A receptor, type 1	Shore et al (2006)	
HyperCKemia, idiopathic	5.21	AD	123320	3p25.3	***CAV3*** *601253*	Caveolin-3	Carbone et al (2000)	allelic to LGMD1C (group 1), MPDT (group 4), RMD2 (group 6), CMH and LQT9 (group 10)
X-linked myopathy with postural muscle atrophy	5.22	XR	XMPMA 300696	Xq26.3	***FHL*** *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	5.23	XD	SPM 300695	Xq26.3	***FHL1*** *300163*	Four-and-a-half LIM domains 1	Quinzii et al (2008)	allelic to EDMD6 (group 1), RBMX1A/B (group 5), XMPMA (group 5)
Reducing body myopathy	5.24	XD	RBMX1A 300717 RBMX1B 300718	Xq26.3	***FHL1*** *300163*	Four-and-a-half LIM domains 1	Schessl et al (2008), Shalaby et al (2009)	allelic to EDMD6 (group 1), XPMA (group 5), SPM (group 5)
Episodic muscle weakness, X-linked	5.25	XR	EMWX 300211	Xp22.3	*?*		Ryan et al (1999)	
Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia	5.26	AD	IBMPFD1 167320	9p13.3	***VCP*** *601023*	Valosin-containing protein	Watts et al (2004) Haubenberger et al (2005)	allelic to scapuloperoneal myopathy (group 1), IBMPFD (group 4), ALS14 (group 12) and CMT2Y (group 14)
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia	5.27		IBMPFD2 615422	7p15.2	***HNRNPA2B1*** *600124*	Heterogeneous nuclear ribonucleoprotein A2/B1	Kim et al (2013)	
Myopathy with exercise intolerance, Swedish type	5.28	AR	HML 255125	12q23.3	***ISCU*** *611911*	Iron-sulfur cluster scaffold homolog (E. coli)	Mochel et al (2008)	
Late onset axial myopathy related to RYR1	5.29	AD		19q13.2	***RYR1*** *180901*	Ryanodine receptor 1 (skeletal)	Løseth et al (2013)	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Tubular aggregate myopathy 1	5.30	AD	TAM1 160565	11p15.4	***STIM1*** *605921*	Stromal interaction molecule 1	Bohm et al (2013) Hedberg et al (2014) Nesin et al (2014)	
Tubular aggregate myopathy 2	5.31	AD	TAM2 615883	12q24.31	***ORAI1*** *610277*	ORAI calcium release-activated calcium modulator 1	Nesin et al (2014)	
Vacuolar myopathy with accumulation of sarcoplasmic reticulum protein aggegates	5.32	AD	VMCQA 616231	1q23.2	***CASQ1*** *114250*	Calsequestrin 1	Rossi et al (2014)	
GROUP 6. MYOTONIC SYNDROMES								
Myotonic dystrophy (Steinert)	6.1	AD	DM1 160900	19q13.32	***DMPK*** *605377*	Dystrophia myotonica protein kinase	Renwick et al (1971) Friedrich et al (1987) Harley et al (1992) Buxton et al (1992) Aslanidis et al (1992) Mahadevan et al (1992) Fu et al (1992) Brook et al (1992)	

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Myotonic dystrophy type 2 (proximal myotonic myopathy)	6.2	AD	DM2 (PROMM) 602668	3q21.3	***CNBP*** *(formerly ZNF9) 116955*	CCHC-type Zinc finger nucleic acid-binding protein (Zinc finger protein 9)	Ranum et al (1998) Liquori et al (2001)	
Myotonia, dominant (Thomsen)	6.3	AD	*see under Ion channel muscle diseases (group 7)*					
Myotonia, recessive (Becker)	6.4	AR	*see under Ion channel muscle diseases (group 7)*					
Rippling muscle disease, dominant	6.5	AD	RMD1 600332	1q41	*?*		Stephan et al (1994)	
Rippling muscle disease, dominant	6.6	AD	RMD2 606072	3p25.3	***CAV3*** *601253*	Caveolin-3	Betz et al (2001)	allelic to LGMD1C (group 1), MPDT (group 4), hyper CKemia (group 5), CMH and LQT9 (group 10)
Rippling muscle disease, recessive	6.7	AR	RMD2 606072	3p25.3	***CAV3*** *601253*	Caveolin-3	Kubisch et al (2003, 2005)	allelic to LGMD1C (group 1), MPDT (group 4), hyper CKemia (group 5), CMH and LQT9 (group 10)
Schwartz-Jampel syndrome	6.8	AR	SJS1 255800	1p36.12	***HSPG2*** *14246*	Heparan sulfate proteoglycan of basement membrane (perlecan)	Nicole et al (1995, 2000)	
Brody disease	6.9	AR	601003	16p11.2	***ATP2A1*** *(formerly SERCA1) 108730*	ATPase, Ca^{++} transporting, fast twitch 1	Odermatt et al, 1996	
Group 7. ION CHANNEL MUSCLE DISEASES								
Chloride channel								
Myotonia congenita, dominant (Thomsen)	7.1	AD	THD 160800	7q34	***CLCN1*** *118425*	Muscle chloride channel	Koch et al. (1992) George et al (1993)	allelic to Becker myotonia (group 7)
Myotonia, recessive (Becker)	7.2	AR	255700	7q34	***CLCN1*** *118425*	Muscle chloride channel	Koch et al (1992)	allelic to Thomsen myotonia (group 7)
Sodium channel								
Hyperkalaemic periodic paralysis	7.3	AD	HYPP 170500	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Fontaine et al (1990) Ptáček et al (1991) Rojas et al (1991) Miller et al (2004)	allelic to HOKPP2, PMC and K-aggravated myotonia (group 7), CMS16 (group 11)
Hypokalaemic periodic Paralysis, type 2	7.4	AD	HOKPP2 613345	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Bulman et al (1999) Jurkat-Rott et al (2000)	allelic to HYPP, PMC and K-aggravated myotonia (group 7), CMS16 (group 11)
Paramyotonia congenita	7.5	AD	PMC 168300	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Ptáček et al (1991-1993) Ebers et al (1991) Koch et al (1992) Mc Clatchey et al (1992)	allelic to HYPP, HOKPP2 and K-aggravated myotonia (group 7), CMS16 (group 11)
Potassium-aggravated myotonia	7.6	AD	608390	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subunit	Ptáček et al (1992, 1994) Heine et al (1993) Lerche et al (1993)	allelic to HYPP, HOKPP2 and PMC (group 7), CMS16 (group 11)
Calcium channel								
Hypokalaemic periodic paralysis, type 1	7.7	AD	HOKPP1 170400	1q32.1	***CACNA1S*** *114208*	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Fontaine et al (1994) Ptáček et al (1994) Jurkat-Rott et al (1994) Elbaz et al (1995)	allelic to MHS5 (group 8)
Acetazolamide responsive hereditary paroxysmal cerebellar ataxia (also listed in group 13 "Ataxias")	7.8	AD	APCA 108500	19p13.13	***CACNA1A*** *601011*	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	von Brederlow et al (1995) Vahedi et al (1995)	allelic to EA2 (group 7), SCA6 (group 13)
Episodic ataxia type-2	7.9	AD	EA2 108500	19p13.13	***CACNA1A*** *601011*	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	Ophoff et al (1996) Jodice et al (1997)	allelic to APCA (group 7), SCA6 (group 13)
Potassium channel								
Hypokalaemic periodic paralysis	7.10	AD	HOKPP 170400	11q13.4	***KCNE3*** *604433*	Potassium channel, voltage-gated, Isk-related family, member 3	Abbott et al (2001)	
Episodic ataxia/myokymia syndrome	7.11	AD	EA1 160120	12p13.32	***KCNA1*** *176260*	Potassium channel, voltage-gated, shaker-related subfamily, member 1	Browne et al. (1994) Adelman et al (1995)	
Thyrotoxic hypokalemic periodic paralysis	7.12		TTPP2 613239	17p11.2	***KCNJ18*** *613236*	Potassium channel, inwardly rectifying, subfamily J, member 18 (Kir2.6)	Ryan et al (2010)	

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Periodic paralysis, potassium sensitive cardiodysrythmic (Andersen's syndrome)	7.13	*see LQ7 under hereditary cardiomyopathies (group 10B, online only)*						
Long QT syndromes	7.14	*see under hereditary cardiomyopathies (group 10B, online only)*						
GROUP 8. MALIGNANT HYPERTHERMIAS								
Malignant hyperthermia	8.1	AD	MHS1 145600	19q13.2	***RYR1*** *180901*	Ryanodine receptor 1 (skeletal)	MacLennan et al (1990) McCarthy et al (1990) Fujii et al (1991) Gillard et al (1991, 1992) Quane et al (1993, 1994) Keating et al (1994	allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), CNMDU1 (group 3)
Malignant hyperthermia	8.2	AD	MHS2 154275	17q11.2-q24	?		Levitt et al. (1992) Moslehi et al (1998)	
Malignant hyperthermia	8.3	AD	MHS3 154276	7q21-q22	?		Iles et al (1994)	
Malignant hyperthermia	8.4	AD	MHS4 600467	3q13.1	?		Sudbrak et al (1995)	
Malignant hyperthermia	8.5	AD	MHS5 601887	1q32.1	***CACNA1S*** *114208*	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Monnier et al (1997)	allelic to HOKPP1 (group 7)
Malignant hyperthermia	8.6	AD	MHS6 601888	5p	?		Robinson et al (1997)	
GROUP 9. METABOLIC MYOPATHIES								
Glycogen storage diseases								
Glycogen storage disease type II, Pompe disease (*also listed in group 10)*	9.1	AR	GSD2 232300	17q25.3	***GAA*** *606800*	Glucosidase, alpha acid	Hers et al (1963) Martiniuk et al (1990) Wokke et al (1995)	
Glycogen storage disease type III	9.2	AR	GSD3 232400	1p21.2	***AGL*** *610860*	Amylo-1, 6-glucosidase, 4-alpha-glucano transferase (glycogen debrancher enzyme)	Sheng et al (1996)	
Glycogen storage disease type IV	9.3	AR	GSD4 232500	3p12.2	***GBE1*** *607839*	Glycogen branching enzyme (1,4-α-glucan branching enzyme)	Brown et al (1966) Bao et al (1996) Bruno et al (2004)	
Glycogen storage disease type V (McArdle)	9.4	AR	GSD5 232600	11q13.1	***PYGM*** *608455*	Glycogen phosphorylase, muscle	Mommaerts et al (1959) Schmidt et al (1959) Lebo et al (1984) Tsujino et al (1993)	
Glycogen storage type VII (Tarui)	9.5	AR	GSD7 232800	12q13.11	***PFKM*** *610681*	Phosphofructokinase, muscle type	Tarui et al (1965) Nakajima et al (1991) Howard et al (1996)	
Glycogen storage disease type IXd (ex type VIII)	9.6	XR	GSD9D 300559	Xq13.1	***PHKA1*** *311870*	Phosphorylase kinase, alpha-1 subunit	Wehner et al (1994) Burwinkel et al (2004)	
Glycogen storage disease type XIV (Congenital disorder of glycosylation, type It)	9.7	AR	CDG1T 614921	1p31.3	***PGM1*** *171900*	Phosphoglucomutase 1	Stojkovic et al (2009)	
Glycogen storage disease type XV	9.8	AR	GSD15 613507	3q24	***GYG1*** *603942*	Glycogenin 1	Moslemi et al (2010)	allelic to PGBM2 (group 9)
Glycogen storage disease type 0	9.9	AR	GSD0B 611556	9q13.33	***GYS1*** *138570*	Glycogen synthase 1	Kolberg et al (2007)	
Glycogen storage disease of heart, lethal congenital	9.10	AD	261740	7q36.1	***PRKAG2*** *602743*	Protein kinase, AMP-activated, non catalytic (AMPK-gamma-2)	Burwinkel et al. (2005)	allelic to CMH6 and WPW syndrome (group 10)
Polyglucosan Body Myopathy 1 with or without immunodeficiency	9.11	AR	PGBM1 615895	20p13	***RBCK1*** *610924*	RanBP-type and C3HC4-type zinc finger containing 1	Nilsson et al (2013)	
Polyglucosan Body Myopathy 2	9.12	AR	PGBM2 616199	3q24	***GYG1*** *603942*	Glycogenin 1	Malfatti et al (2014)	allelic to GSD15 (group 9)
Glycolytic pathway								
Posphoglycerate kinase 1 deficiency	9.13	XR	300653	Xq21.1	***PGK1*** *311800*	Phosphoglycerate kinase 1	DiMauro et al (1981, 1983) Rosa et al (1982)	
Glycogen storage disease type X	9.14	AR	GSD10 261670	7p13	***PGAM2*** *612931*	Phosphoglycerate mutase 2	DiMauro et al. (1981) Edwards et al. (1989) Castella-Escola et al (1990) Tsujino et al. (1993)	
Glycogen storage disease type XI	9.15	AR	GSD11 612933	11p15.1	***LDHA*** *150000*	Lactate dehydrogenase A	Kanno et al (1980) Scrable et al (1990)	

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Glycogen storage disease type XIII	9.16	AD	GSD13 612932	17p13.2	***ENO3*** *131370*	Enolase 3	Comi et al (2001)	
Erythrocyte lactate transporter defect (Myopathy due to actate transporter defect)	9.17	AD	245340	1.13.2	***SLC16A*** *600682*	Solute carrier family 16 (monocarboxylic acid transporter), member 1	Merezhinskaya et al (2000)	
Disorders of lipid metabolism								
Carnitine palmitoyl-transferase II deficiency, myopathic, stress induced	9.18	AR	255110	1p32.3	***CPT2*** *600650*	Carnitine palmitoyl transferase II	DiMauro et al (1973) Finocchiaro et al (1991) Taroni et al (1993) Gellera et al (1994)	
Primary systemic carnitine deficiency	9.19	AR	CDSP 212140	5q31.1	***SLC22A5*** *603377*	Solute carrier family 22 (organic cation transporter), member 5	Nezu et al (1999)	
Carnitine/acyl-carnitine translocase deficiency	9.20	AR	CACTD 212138	3p21.31	***SLC25A20*** *613698*	Solute carrier family 25 (carnitine/ acylcarnitine translocase), member 20	Huizing et al (1997) Ogawa et al (2000)	
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIA)	9.21	AR	MADD 231680	15q24.2-q24.3	***ETFA*** *608053*	Electron-transfer-flavoprotein, alpha polypeptide	Indo et al (1991) Freneaux et al (1992)	allelic to MADD-GaIIB and GaIIC (group 9)
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIB)	9.22	AR	MADD 231680	19q13.41	***ETFB*** *130410*	Electron-transfer-flavoprotein, beta polypeptide	Colombo et al (1994)	allelic to MADD-GaIIA and GaIIC (group 9)
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIC, riboflavin responsive)	9.23	AR	MADD 231680	4q32.1	***ETFDH*** *231675*	Electron-transfer-flavoprotein dehydrogenase	Beard et al (1993)	allelic to MADD-GaIIA and GaIIB (group 9)
Acyl-CoA dehydrogenase (very long chain) deficiency (VLCAD deficiency)	9.24	AR	ACADVLD 201475	17p13.1	***ACADVL*** *609575*	Acyl-Coenzyme A dehydrogenase, very long chain	Aoyama (1993, 1995) Strauss et al (1995) Mathur et al 1999)	
Mitochondrial complex I deficiency due to ACAD9 deficiency	9.25	AR	611126	3q21.3	***ACAD9*** *611103*	Acyl-CoA dehydrogenase family member 9 (**M**)	Fragaki etal (2016)	
Triglyceride storage disease with ichthyosis (Chanarin-Dorfman syndrome)	9.26	AR	CDS 275630	3p21.33	***ABHD5*** *604780*	Abhydrolase domain containing 5	Lefevre et al (2001)	
Neutral lipid storage disease with myopathy without ichthyosis	9.27	AR	NLSDM 610717	11p15.5	***PNPLA2*** *609059*	Patatin-like phosphorylase domain-containing protein 2 (Adipose triglyceride lipase)	Fischer et al (2007)	
Acute Recurrent myoglobinuria	9.28	AR	268200	2p25.1	***LPIN1*** *605518*	lipin 1 (phsosphatidic acid phosphatase 1)	Zeharia et al (2008)	
Mitochondrial myopathy with lactic acidosis	9.29	AR	MMLA 251950	7q31.1	***PNPLA8*** *612123*	Patatin-like phospholipase domain containing 8 (**M**)	Saunders et al (2015)	
Lipid storage myopathy due to Flavin adenine dinucleotide synthetase deficiency	9.30	AR	LSMFLAD 255100	1q21.3	***FLAD1*** *610595*	Flavin adenine dinucleotide synthetase, S. Cerevisiae, homolog of (**M**)	Taylor et al (2014)	
GROUP 10A. HEREDITARY CARDIOMYOPATHIES non arrhythmogenic								
Hypertrophic cardiomyopathies								
Familial hypertrophic cardiomyopathy, 1	10.1	AD	CMH1 192600	14q11.2	***MYH7*** *160760*	Myosin heavy chain 7 (beta), cardiac muscle	Jarcho et al (1989) Solomon et al (1990) Tanigawa et al (1990) Geisterfer-Lowrance et al (1990)	allelic to CFTD and MSMB (group 3), MPD1 (group 4), CMD1S and LVNC5 (group 10)
Familial hypertrophic cardiomyopathy, 2	10.2	AD	CMH2 115195	1q32.1	***TNNT2*** *191045*	Cardiac troponin T	Watkins et al (1993) Thierfelder et al (1994)	allelic to CMD1D, RSM3 and and LVNC6 (group 10)
Familial hypertrophic cardiomyopathy, 3	10.3	AD	CMH3 115196	15q22.2	***TPM1*** *191010*	Tropomyosin-1	Thierfelder et al (1994)	allelic to CMD1Y and LVNC9 (group 10)
Familial hypertrophic cardiomyopathy, 4	10.4	AD	CMH4 115197	11p11.2	***MYBPC3*** *600958*	Cardiac myosin binding protein-C	Carrier et al (1993) Bonne et al (1995) Watkins et al (1995)	allelic to congenital skeletal myopathy and fatal cardiomyopathy (group 3), CMD1MM and LVNC10 (group 10)

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Familial hypertrophic cardiomyopathy, 6	10.5	AD	CMH6 600858	7q36.1	***PRKAG2*** *602743*	Protein kinase, AMP-activated, non catalytic (AMPK-gamma-2)	Blair et al (2001)	allelic to glycogen storage disease of heart, lethal congenital (group 9)
Familial hypertrophic cardiomyopathy, 7	10.6	AD	CMH7 613690	19q13.42	***TNNI3*** *191044*	Cardiac troponin I	Kimura et al (1997)	allelic to RCM1, CMD1FF and CMD2A (group 10)
Familial hypertrophic cardiomyopathy, 8	10.7	AD	CMH8 608751	3p21.31	***MYL3*** *160790*	Myosin, light chain 3, alkali; ventricular, skeletal, slow	Poetter et al (1996)	
Familial hypertrophic cardiomyopathy, 9	10.8	AD	CMH9 613765	2q31.2	***TTN*** *188840*	Titin	Satoh et al (1999)	allelic to LGMD2J (group 1), SALMY (group 3), TMD (group 4), HMERF (group 5), CMD1G (group 10)
Familial hypertrophic cardiomyopathy, 10	10.9	AD	CMH10 608758	12q24.11	***MYL2*** *160781*	Myosin, light chain 2, regulatory, cardiac, slow	Poetter et al (1996)	
Familial hypertrophic cardiomyopathy, 11	10.10	AD	CMH11 612098	15q14	***ACTC1*** *102540*	Actin, alpha, cardiac muscle	Mogensen et al (1999)	alllelic to CMD1R, LVNC4 (group 10)
Familial hypertrophic cardiomyopathy, 12	10.11	AD	CMH12 612124	11p15.1	***CSRP3*** *600824*	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Geier et al (2008)	allelic to CMD1M (group 10)
Familial hypertrophic cardiomyopathy, 13	10.12	AD	CMH13 613243	3p21.1	***TNNC1*** *191040*	Slow troponin C	Landstrom et al (2008)	allelic to CMD1Z (group 10)
Familial hypertrophic cardiomyopathy, 14	10.13	AD	CMH14 613251	14q11.2	***MYH6*** *160710*	Myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al (2005)	allelic to CMD1EE and SSS3 (group 10)
Hypertrophic cardiomyopathy, 15	10.14	AD	CMH15 613255	10q22.2	***VCL*** *193065*	Vinculin	Vasile et al (2006)	allelic to CMD1W (group 10)
Hypertrophic cardiomyopathy	10.15	AD, digenic	CMH1 192600	20q11.21	***MYLK2*** *606566*	Myosin light chain kinase 2	Davis et al (2001)	
Hypertrophic cardiomyopathy	10.16	AD	CMH1 192600	3p25.3	***CAV3*** *601253*	Caveolin-3	Hayashi et al (2004) Fulizio et al (2005)	allelic to LGMD1C (group 1), hyperCKemia (group 5), MPDT (group 4), RMD2 (group 6)
Hypertrophic cardiomyopathy, 16	10.17	AD	CMH16 613838	4q26	***MYOZ2*** *605602*	Myozenin 2 (calsarcin 1)	Osio et al (2007)	
Hypertrophic cardiomyopathy, 17	10.18	AD	CMH17 613873	20q13.12	***JPH2*** *605267*	Junctophilin-2	Landstrom et al (2007) Matsuhita et al (2007)	
Hypertrophic cardiomyopathy, 18	10.19	AD	CMH18 613874	6q22.31	***PLN*** *172405*	Phospholamban	Minamisawa et al (2003) Landstrom et al (2011)	allelic to CMD1P (group 10)
Hypertrophic cardiomyopathy, 19	10.20	AD	CMH19 61387	19p13.11	***CALR3*** *611414*	Calreticulin 3	Chiu et al (2007)	
Hypertrophic cardiomyopathy, 20	10.21	AD	CMH20 613876	1p31.1	***NEXN*** *613121*	Nexilin F-actin binding protein	Wang et al (2010)	allelic to CMD1CC (group 10)
Hypertrophic cardiomyopathy related to cardiac ankyrin repeat domain protein	10.22	AD		10q23.33	***ANKRD1*** *609599*	Ankyrin repeat domain-protein 1	Arimura et al (2009)	allelic to dilated cardiomyopathy (group 10)
Hypertrophic cardiomyopathy, 22	10.23	AD	CMH22 615248	10q21.3	***MYPN*** *608517*	Myopalladin	Purevjav et al (2012)	allelic to CMD1KK (group 10) and NEM11 (group 3)
Hypertrophic cardiomyopathy, 23	10.24	AD	CMH23 612158	1q43	***ACTN2*** *102573*	Actinin alpha-2	Chiu C et al (2010)	allelic to CMD1AA (group 10)
Hypertrophic cardiomyopathy, 24	10.25	AD	CMH24 601493	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Theis et al (2006)	allelic to allelic to allelic to Markesbery-Griggs (group 4), MFM4 (group 5), CMD1C and LVNC3 (group 10)
Hypertrophic cardiomyopathy, 25	10.26	AD	CMH25 607487	17q12	***TCAP*** *604488*	Titin-cap (telethonin)	Hayashi et al (2004)	allelic to LGMD2G (group 1), CMD1N (group 10)
Hypertrophic cardiomyopathy, 26	10.27	AD	CMH26 617047	7q32.1	***FLNC*** *102565*	Filamin C	Valdes-Mas et al (2014)	allelic to MFM5 (group 5) and MPD4 (group 4) and RCM5 (group 10)
Hypertrophic cardiomyopathy, 27	10.28	AR	CMH27 618052	15q25.3	***ALPK3*** *617608*	Alpha Kinase 3	Almomani et al (2016)	
Mitochondrial hypertrophic cardiomyopathy related to *NDUFAF1*	10.29	AR	252010	15q15.1	***NDUFAF1*** *606934*	NADH dehydrogenase (ubiquinone) complex I, Assembly factor 1 (**M**)	Fassone et al (2011)	
Mitochondrial hypertrophic cardiomyopathy related to *TSFM*	10.30	AR	COXPD3 610505	12q14.1	***TSFM*** *604723*	Ts translation elongation factor, mitochondrial (**M**)	Smeitink et al.(2006)	
Mitochondrial hypertrophic cardiomyopathy related to *AARS2*	10.31	AR	COXPD8 614096	6p21.1	***AARS2*** *612035*	Alanyl-tRNA synthetase 2, mitochondrial (**M**)	Götz et al (2011)	

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Mitochondrial hypertrophic cardiomyopathy related to *MRPL3*	10.32	AR	COXPD9 614582	3q21.1	***MRPL3*** *607118*	Mitochondrial ribosomal protein L3 (**M**)	Galmiche et al (2011)	
Mitochondrial hypertrophic cardiomyopathy related to *MTO1*	10.33	AR	COXPD10 614702	6q13	***MTO1*** *614667*	Mitochondrial tRNA translation optimization 1 (**M**)	Ghezzi et al (2012) Baruffini et al (2013)	
Mitochondrial hypertrophic cardiomyopathy related to *MRPL44*	10.34	AR	COXPD16 615395	2q36.1	***MRPL44*** *611849*	Mitochondrial ribosomal protein L44 (**M**)	Carroll et al (2013)	
Cardio-encephalo-myopathy, fatal infantile, related to *SCO2*	10.35	AR	CEMCOX1 604377	22q13.33	***SCO2*** *604272*	Cytochrome c oxidase assembly protein (**M**)	Papadopoulou et al (1999)	allelic to CMT2 related to SCO2
Mitochondrial hypertrophic cardiomyopathy, related to COX15	10.36	AR	CEMCOX2 615119	10q24.2	***COX15*** *603646*	Cytochrome c oxidase assembly factor Cox15 (**M**)	Antonicka et al (2003)	
Dilated cardiomyopathies								
Dilated cardiomyopathy, 1A	10.37	AD	CMD1A 115200	1q22	***LMNA*** *150330*	Lamin A/C	Fatkin et al (1999)	allelic to EDMD2, EDMD3 and LGMD1B (group1), MDCL (group 2), CMT2B1 (group 14) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
Dilated cardiomyopathy, 1B	10.38	AD	CMD1B 600884	9q13	*?*		Krajinovic et al (1995)	
Dilated cardiomyopathy, 1C	10.39	AD	CMD1C 601493	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Bowles et al (1996) Vatta et al (2003) Arimura et al (2004)	allelic to allelic to Markesbery-Griggs (group 4), MFM4 (group 5), CMH24 and LVNC3 (group 10)
Dilated cardiomyopathy, 1D	10.40	AD	CMD1D 601494	1q32.1	***TNNT2*** *191045*	Troponin type 2 (cardiac)	Durand et al (1995) Kamisago et al (2000)	allelic to CMH2, RSM3 and and LVNC6 (group 10)
Dilated cardiomyopathy, 1E	10.41	AD	CMD1E 601154	3p22.2	***SCN5A*** *600163*	Sodium channel, voltage-gated, type V, alpha	McNair et al (2004)	allelic to ATFB10, BRGDA1, LQT3, SSS1 (group 10)
Dilated cardiomyopathy, 1G	10.42	AD	CMD1G 604145	2q31.2	***TTN*** *188840*	Titin	Siu et al (1999) Gerull et al (2002) Itoh-Satoh et al (2002)	allelic to LGMD2J (group 1), SALMY (group 3), TMD (group 4), HMERF (group 5), CMH9 (group 10),
Dilated cardiomyopathy, 1H	10.43	AD	CMD1H 604288	2q14-q22	*?*		Jung et al (1999)	
Dilated cardiomyopathy, 1I	10.44	AD	CMD1I 604765	2q35	***DES*** *125660*	Desmin	Li et al (1999)	allelic to LGMD1E and LGM2R (group 1), ARVD7 (group 10)
Dilated cardiomyopathy, 1J	10.45	AD	CMD1J 605362	6q23.2	***EYA4*** *603550*	Eyes absent homolog	Schönberger et al (2005)	
Dilated cardiomyopathy, 1K	10.46	AD	CMD1K 605582	6q12-q16	*?*		Sylvius et al (2001)	
Dilated cardiomyopathy, 1L	10.47	AD	CMD1L 606685	5q33.2-q33.3	***SGCD*** *601411*	Sarcoglycan, delta	Tsubata et al (2000)	allelic to LGMD2F (group 1)
Dilated cardiomyopathy, 1M	10.48	AD	CMD1M 607482	11p15.1	***CSRP3*** *600824*	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Knoll et al (2002)	allelic to CMH12 (group 10)
Dilated cardiomyopathy, 1N	10.49	AD	CMD1N 607487	17q12	***TCAP*** *604488*	Titin-cap (Telethonin)	Knoll et al (2002)	allelic to LGMD2G (group 1), CMH25 (group 10)
Dilated cardiomyopathy, 1O	10.50	AD	CMD1O 605569	12p12.1	***ABCC9*** *601439*	ATP-binding cassette, subfamily C, member 9	Bienengraeber etal (2004)	allelic to ATFB12 (group 10)
Dilated cardiomyopathy, 1P	10.51	AD	CMD1P 609909	6q22.31	***PLN*** *172405*	phospholamban	Schmitt et al (2003) Haghighi et al (2003, 2006)	allelic to CMH18 (group 10)
Dilated cardiomyopathy, 1Q	10.52	AD	CMD1Q 609915	7q22.3-q31.1	*?*		Schonberger et al (2005)	
Dilated cardiomyopathy, 1R	10.53	AD	CMD1R 613424	15q14	***ACTC1*** *102540*	actin, alpha, cardiac muscle	Olson et al (1998) Mogensen et al (1999)	allelic to CMH11, LVNC4 (group 10)
Dilated cardiomyopathy, 1S	10.54	AD	CMD1S 613426	14q11.2	***MYH7*** *160760*	Myosin heavy chain 7 (beta), cardiac muscle	Kamisago et al (2000)	allelic to CFTD and MSMB (group 3), MPD1 (group 4), CMH1 and LVNC5 (group 10)
Dilated cardiomyopathy, 1T	10.55	AD	CMD1T	12q23.1	***TMPO*** *188380*	Thymopoietin (lamina-associated polypeptide 2)	Taylor et al (2005)	
Dilated cardiomyopathy, 1U	10.56	AD	CMD1U 613694	14q24.2	***PSEN1*** *104311*	Presenilin 1	Li et al (2006)	
Dilated cardiomyopathy, 1U	10.57	AD	CMD1V 613697	1q42.13	***PSEN2*** *600759*	Presenilin 2	Li et al (2006)	

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Dilated cardiomyopathy, 1W	10.58	AD	CMD1W 611407	10q22.2	***VCL*** *193065*	Vinculin	Olson et al (2002) Vasile et al (2006)	allelic to CMH15 (group 10)
Dilated cardiomyopathy related, 1X	10.59	AR	CMD1X 611615	9q31.2	***FKTN*** *607440*	Fukutin	Murakami et al (2006)	allelic to LGMD2M (group 1), FCMD and WWS (group 2)
Dilated cardiomyopathy, 1Y	10.60	AD	CMD1Y 611878	15q22.2	***TPM1*** *191010*	Tropomyosin-1	Olson et al (2010)	allelic to CMH3 and LVNC9 (group 10)
Dilated cardiomyopathy, 1Z	10.61	AD	CMD1Z 611879	3p21.1	***TNNC1*** *191040*	Slow troponin C	Mogensen et al (2004)	allelic to CMH13 (group 10)
Dilated cardiomyopathy, 1AA	10.62	AD	CMD1AA 612158	1q42-q43	***ACTN2*** *102573*	Actinin alpha-2	Mohapatra et al (2003)	allelic to CMH23 (group 10)
Dilated cardiomyopathy, 1BB	10.63	AD/AR	CMD1BB 612877	18q12.1	***DSG2*** *125671*	Desmoglein 2	Posch et al (2008)	allelic to ARVD10 (group 10)
Dilated cardiomyopathy, 1CC	10.64	AD	CMD1CC 613122	1p31.1	***NEXN*** *613121*	Nexilin F-actin binding protein	Hassel et al (2009)	allelic to CMH20 (group 10)
Dilated cardiomyopathy, 1DD	10.65	AD	CMD1DD 613172	10q25.2	***RBM20*** *613171*	RNA-binding motif protein 20	Brauch et al (2009)	
Dilated cardiomyopathy, 1EE	10.66	AD	CMD1EE 613252	14q11.2	***MYH6*** *160710*	Myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al (2005)	allelic to CMH14 (group 10)
Dilated cardiomyopathy, 1FF	10.67	AD	CMD1FF 613286	19q13.42	***TNNI3*** ***191044***	Cardic troponin I	Carballo et al (2009)	allelic to CMH7, CDM2A, RCM1 (group 10)
Dilated cardiomyopathy, 1GG	10.68	AR	CMD1GG 613642	5p15.33	***SDHA*** *600857*	Succinate dehydrogenase complex, subunit a, flavoprotein (**M**)	Levitas et al (2010)	
Dilated cardiomyopathy, 1HH	10.69	AD	CMD1HH 613881	10q26.11	***BAG3*** *603883*	Bcl2-associated athanogene 3	Norton et al (2011)	allelic to MMF6 (group 5) and CMT2 (group 14)
Dilated cardiomyopathy, 1II	10.70	AD	CMD1II 615184	11q23.1	***CRYAB*** *123590*	Alpha-B crystallin	Inagaki et al (2006)	allelic to MFM2 (group 5)
Dilated cardiomyopathy, 1JJ	10.71	AD	CMD1JJ 615235	6q21	***LAMA4*** *600133*	Laminin, alpha-4	Knöll et al (2007)	
Dilated cardiomyopathy, 1KK	10.72	AD	CMD1KK 615248	10q21.3	***MYPN*** *608517*	Myopalladin	Duboscq-Bidot (2008)	allelic to NEM11 (group 3) and CMH22 (group 10)
Dilated cardiomyopathy, 1LL	10.73	AD	CMD1LL 615373	1p36.32	***PRDM16*** *605557*	PR domain-containing protein 16	Arndt et al (2013)	allelic to LVNC8 (group 10)
Dilated cardiomyopathy, 1MM	10.74	AD	CMD1MM 615396	11p11.2	***MYBPC3*** *600958*	Myosin-binding protein C, cardiac	Hershberger et al (2010)	allelic to congenital myopathy and fatal cardiomyopathy (group 3), CMH4 and LVNC10 (group 10)
Dilated cardiomyopathy, 1NN	10.75	AD	CMD1NN 615916	3p25.2	***RAF1*** *164760*	V-Raf-1 murine leukemia viral oncogene homolog 1	Dhandapany et al (2014)	
Dilated cardiomyopathy related to integrin-linked kinase	10.76	AD		11p15.4	***ILK*** *602366*	Integrin-linked kinase	Knöll et al (2007)	
Dilated cardiomyopathy related to cardiac ankyrin repeat protein	10.77	AD		10q23.31	***ANKRD1*** *609599*	Ankyrin repeat domain 1 containing protein1	Duboscq-Bidot et al (2009) Moulik et al (2009)	
Dilated cardiomyopathy, 2A	10.78	AR	CMD2A 611880	19q13.42	***TNNI3*** *191044*	Cardiac troponin I	Carballo et al (2009)	allelic to CMH7, CMD1FF, RCM1 (group 10)
Dilated cardiomyopathy 2B	10.79	AR	CMD2B 614672	7q21.2	***GATAD1*** *614518*	GATA zinc finger domain-containing protein 1	Theis et al (2011)	
Dilated cardiomyopathy, 3A	10.80	XR	BTHS 302060	Xq28	***TAZ*** *300394*	Tafazzin	Gedeon et al (1995)	allelic to BTHS (group 10)
Dilated cardiomyopathy, 3B	10.81	XR	CMD3B 302045	Xp21.2-p21.1	***DMD*** *300377*	Dystrophin	Muntoni et al (1993) Milasin et al (1996)	allelic to DMD and BMD (group 1)
Dilated cardiomyopathy related to nesprin-1	10.82	AD		6q25.2	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope protein 1 (nesprin-1)	Puckelwartz et al (2010)	allelic to EDMD4 (group 1) SCAR8 (group 13), AMC with nesprin-1 defect (group 16)
Dilated cardiomyopathy related to MURC	10.83	AD		9q31.1	***CAVIN4*** *617714*	Caveolae-associated protein 4 (MURC)	Rodriguez et al (2011)	
Dilated cardiomyopathy related to *DOLK*	10.84	AR	CDGM1 610768	9q34.11	***DOLK*** *610746*	Dolichol kinase	Kranz et al (2007) Lefeber et al (2011)	
Restrictive cardiomyopathies								
Restrictive cardiomyopathy, 1	10.85	AD	RCM1 115210	19q13.42	***TNNI3*** *191044*	Cardiac troponin I	Mogensen et al (2003)	allelic to CMH7, CMD1FF, CMD2A (group 10)
Restrictive cardiomyopathy, 2	10.86		RCM2 609578	10q23.3	*?*		Zhang et al (2005)	

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Restrictive cardiomyopathy, 3	10.87	AD	RCM3 612422	1q32.1	***TNNT2*** *191045*	Cardiac Troponin T	Peddy et al (2006)	allelic to CMH2, CMD1D and LVNC6 (group 10)
Restrictive cardiomyopathy, 4	10.88	AD	RCM4 615248	10q21.3	***MYPN*** *608517*	Myopalladin	Purevjav et al (2012)	allelic to CMH22 (group 10), CMD1KK (group 10) and NEM11 (group 3)
Restrictive cardiomyopathy, 5	10.89	AD	RCM5 617047	7q32.1	***FLNC*** *102565*	Filamin C	Brodehl et al (2016)	allelic to MFM5 (group 5), MPD4 (group 4) and CMH26 (group 10)
Other non arrhythmogenic hereditary cardiomyopathies								
Pompe disease, Glycogenosis, generalized, cardiac form (*also listed in group 9*)	10.90	AR	GSDII 232300	17q25.3	***GAA*** *606800*	Glucosidase, alpha; acid	Hers (1963) Martiniuk et al (1990) Wokke et al (1995)	
Cardioskeletal myopathy with neutropenia and abnormal mitochondria (Barth syndrome)	10.91	XR	BTHS 302060	Xq28	***TAZ*** *300394*	Tafazzin	Bolhuis et al (1991) Bione et al (1996)	allelic to CMD3A (group 10)
Left ventricular noncompaction, 1	10.92	AD	LVNC1 604169	18q12.1	***DTNA*** *601239*	Dystrobrevin, alpha	Ichida et al (2001)	
Left ventricular noncompaction, 2	10.93		LVNC2 609470	11q15	***?***		Sasse-Klaassen et al (2004)	
Left ventricular noncompaction, 3	10.94	AD	LVNC3 601493	10q23.2	***LDB3*** *605906*	LIM domain binding-3	Vatta et al (2003)	alllelic to Markesbery-Griggs (group 4), MFM4 (group 5), CMH24, CMD1C (group 10),
Left ventricular noncompaction, 4	10.95	AD	LVNC4 613424	15q14	***ACTA1*** *102540*	Actin, alpha, cardiac muscle	Monserrat et al (2007)	allelic to CMH11, CMD1R (group 10)
Left ventricular noncompaction, 5	10.96	AD	LVNC5 613426	14q11.2	***MYH7*** *160760*	Myosin heavy chain 7 (beta), cardiac muscle	Klaassen et al (2008)	allelic to CFTD and MSMB (group 3), MPD1 (group 4), CMH1 and CMD1S (group 10)
Left ventricular noncompaction, 6	10.97	AD	LVNC6 601494	1q32.1	***TNNT2*** *191045*	Cardiac Troponin T	Luedde et al (2010)	allelic to CMH2, CMD1D and RCM3 (group 10)
Left ventricular noncompaction, 7	10.98	AD	LVNC7 615092	18q11.2	***MIB1*** *608677*	Mindbomb, drosophila, homolog of	Luxan et al. (2013)	
Left ventricular noncompaction, 8	10.99	AD	LVNC8 615373	1p36.32	***PRDM16*** *605557*	PR domain-containing protein 16	Arndt et al. (2013)	allelic to CMD1LL (group 10)
Left ventricular noncompaction, 9	10.100	AD	LVNC9 611878	15q22.2	***TPM1*** *191010*	Tropomyosin-1	Probst et al. (2011)	allelic to CMH3, CMD1Y (group 10)
Left ventricular noncompaction, 10	10.101	AD	LVNC10 615396	11p11.2	***MYBPC3*** *600958*	Cardiac myosin binding protein-C	Probst et al. (2011)	allelic to congenital skeletal myopathy and fatal cardiomyopathy (group 3), CMH4 and CMD1MM (group 10)
Cardiovalvular dysplasia, X-linked (Myxomatous valvular dystrophy)	10.102	XR	CVD1 314400	Xq28	***FLNA*** *300017*	Filamin A, alpha (actin binding protein 280)	Kyndt et al (1998) Kyndt et al (2007)	
GROUP 10-B. FOR ARRHYTHMOGENIC CARDIOMYOPATHIES AND RELATED SYNDROMES see online version of the gene table http://www.musclegenetable.fr								
GROUP 11. CONGENITAL MYASTHENIC SYNDROMES								
Slow channel syndromes	11.1	AD	CMS1A 601462	2q31.1	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1	Sine et al (1995) Engel et al (1996) Croxen et al (1997)	allelic to other entries in group 11
	11.2	AD	CMS2A 616313	17p13.1	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1	Engel et al (1996b) Gomez et al (1996)	allelic to other entries in group 11
	11.3	AD	CMS3A 616321	2q37.1	***CHRND*** *100720*	Cholinergic receptor, Cicotinic, delta	Gomez et al (2002)	allelic to other entries in group 11
	11.4	AD, AR	CMS4A 605809	17p13.2	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Ohno et al (1995) Gomez et al (1995) Engel et al (1996) Croxen et al (2002)	allelic to other entries in group 11
Fast channel syndromes	11.5	AR	CMS1B 608930	2q31.1	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1	Wang et al (1999) Shen et al (2003)	allelic to other entries in group 11
	11.6	AR	CMS3B 616322	2q37.1	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Brownlow et al (2001)	allelic to other entries in group 11
	11.7	AR	CMS4B 616324	17p13.2	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Ohno et al (1996)	allelic to other entries in group 11
Acetylcholine receptor deficiency	11.8	AR	CMS2C 616314	17p13.1	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1	Quiram et al (1999)	allelic to other entries in group 11
	11.9	AR	CMS3C 616323	2q37.1	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Shen et al (2002)	allelic to other entries in group 11

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	11.10	AR	CMS4C 608931	17p13.2	***CHRNE*** *100725*	Ccholinergic receptor, nicotinic, epsilon	Engel et al (1996) Ohno et al (1997)	allelic to other entries in group 11
Myasthenic syndrome, congenital, 5	11.11	AR	CMS5 603034	3p25.1	***COLQ*** *603033*	Collagenic tail of endplate acetylcholinesterase	Donger et al (1998) Ohno et al (1998-2000)	
Myasthenic syndrome, congenital, 6, presynaptic	11.12	AR	CMS6 254210	10q11.23	***CHAT*** *118490*	Choline acetyltransferase	Ohno et al (2001) Maselli et al (2003)	
Myasthenic syndrome, congenital, 7, presynaptic	11.13	AD	CMS7 616040	1q32.1	***SYT2*** *600104*	Synaptotagmin 2	Herrmann et al (2014)	
Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	11.14	AR	CMS8 615120	1p36.33	***AGRN*** *103320*	Agrin	Huzé et al (2009)	
Myasthenic syndrome, congenital, 9	11.15	AR	CMS9 616325	9q31.3	***MUSK*** *601296*	Muscle-specific receptor tyrosine kinase	Chevessier et al (2004)	allelic to FADS (group 16)
Myasthenic syndrome, congenital, 10	11.16	AR	CMS10 254300	4p16.3	***DOK7*** *610285*	Downstream of tyrosin kinase 7	Beeson et al (2006) Selcen et al (2008)	allelic to FADS (group 16)
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	11.17	AR	CMS11 616326	11p11.2	***RAPSN*** *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	11.18	AR	CMS12 610542	2p13.3	***GFPT1*** *138292*	Glutamine: fructose-6-phosphate amido transtransferase 1	Senderek et al (2011)	
Myasthenic syndrome, congenital, 13, with tubular aggregates	11.19	AR	CMS13 614750	11q23.3	***DPAGT1*** *191350*	Dolichyl-phosphate N-acetyl-glucosamine phos-photransferase 1	Belaya et al (2012)	
Myasthenic syndrome, congenital, 14, with tubular aggregates	11.20	AR	CMS14 616228	9q22.33	***ALG2*** *607905*	S. Cerevisae homolog of ALG2 (alpha-1,3/1,6-mannosyl transferase)	Cossins et al (2013)	
Myasthenic syndrome, congenital, 15, without tubular aggregates	11.21	AR	CMS15 607227	1p21.3	***ALG14*** *612866*	S. Cerevisae homolog of ALG14 (UDP-N-acetyl-glucosami-nyltransferase subunit)	Cossins et al (2013)	
Myasthenic syndrome, congenital, 16	11.22	AR	CMS16 614198	17q23.3	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha subnit	Tsujino et al (2003)	allelic to HOKPP2, HYPP, PMC and K-aggravated myotonia (group 7)
Myasthenic syndrome, congenital, 17	11.23	AR	CMS17 616304	11p11.2	***LRP4*** *604270*	LDL receptor-related protein 4	Ohkawara et al (2014)	
Myasthenic syndrome, congenital, 18	11.24	AD	CMS18 616330	20p12.2	***SNAP25*** *600322*	Synaptosomal associated protein 25	Shen et al (2014)	
Myasthenic syndrome, congenital, 19	11.25	AR	CMS19 616720	10q22.1	***COL13A1*** *120350*	Collagen type XIII alpha 1 chain	Logan et al (2015)	
Myasthenic syndrome, congenital, 20	11.26	AR	CMS20 617143	2q12.3	***SLC5A7*** *608761*	Solute carrier family 5 (choline transporter) member 7	Bauche et al (2016)	allelic to HMN7A (group 12)
Myasthenic syndrome, congenital, 21, presynaptic	11.27	AR	CMS21 617239	10q11.23	***SLC18A3*** *600336*	Solute carrier family 18 (vesicular acetylcholine), member 3	O'Grady et al (2016)	
Myasthenic syndrome, congenital, 22	11.28	AR	CMS22 616224	2p21	***PREPL*** *609557*	Prolyl endopeptidase-like	Regal et al (2014)	
Congenital myasthenic syndrome with nephrotic syndrome	11.29	AR	NPHS5 614199	3p21.31	***LAMB2*** *150325*	Laminin-beta 2	Maselli et al (2009)	
Escobar syndrome (multiple pterygium syndrome)	11.30	AR	EVMPS 265000	2q37.1	***CHRNG*** *100730*	Cholinergic receptor, nicotinic, gamma	Hoffman et al (2006) Morgan et al (2006)	
Myasthenic syndrome, with plectin defect	11.31	AR		8q24.3	***PLEC*** *601282*	Plectin	Banwell et al (1999) Forrest et al (2010) Selcen et al (2011)	Allelic to LGMD2Q (group 1), EBSMD (group 5)
Congenital myasthenia	11.32	AR		15q23	***MYO9A*** *604875*	Myosin IXA	O'Connor et al (2016)	

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Congenital myasthenic syndrome related to *GMPPB*	11.33	AR		3q21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Belaya et al (2015)	allelic to LGMD2T (group 1); MEB, MDDGB14 (group 2)
Presynaptic congenital myasthenic syndrome	11.34	AR		12p13.31	***VAMP1*** *185880*	Vesicle-associated membrane protein 1	Shen et al (2017) Salpieto et al (2017)	allelic to SPAX1 (group 15)
Presynaptic congenital myasthenic syndrome	11.35	AR		20q13.33	***LAMA5*** *601033*	Laminin, alpha-5	Maselli et al (2017)	
Presynaptic congenital myasthenic syndrome related to MUNC13-1	11.36	AR		9p13.3	***UNC13*** *605836*	C. Elegans, homolog of UNC13B (MUNC13)	Engel et al (2016)	
Congenital myasthenic syndrome related to SLC25A1	11.37	AR		22q11.21	***SLC25A1*** *190315*	Solute carrier family 25 (mitochondrial carrier, citrate transporter), member 1	Chaouch et al 2014)	
Congenital myasthenic syndrome related to RPH3A, presynaptic	11.38	AR		12q24.13	***RPH3A*** *612159*	Rabphilin 3A	Maselli et al (2018)	
GROUP 12. SPINAL MUSCULAR ATROPHIES MOTONEURON DISEASES								
Spinal muscular atrophy related to SMN1								
Spinal muscular atrophy, type I (Werdnig-Hoffman)	12.1	AR	SMA1 253300	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Gilliam et al (1990) Melki et al (1990, 1994) Lefebvre et al (1995) Bussaglia et al (1995) Rodrigues et al (1995) Roy et al (1995) Hahnen et al (1997)	allelic to SMA2, SMA3, SMA4 (group 12)
Spinal muscular atrophy, type II (intermediate)	12.2	AR	SMA2 253550	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Matthijs et al (1996) Samilchuk (1996)	allelic to SMA2, SMA3, SMA4 (group 12)
Spinal muscular atrophy, type III (Kugelberg-Welander)	12.3	AR	SMA3 253400	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Brzustowicz et al (1990) Melki et al (1990b) Lefebvre et al (1995)	allelic to SMA2, SMA3, SMA4 (group 12)
Spinal muscular atrophy, type IV, adult form	12.4	AR	SMA4 271150	5q13.2	***SMN1*** *600354*	Survival of motor neuron 1	Brahe et al (1995) Clermont et al (1995)	allelic to SMA2, SMA3, SMA4 (group 12)
Distal spinal muscular atrophy, recessive								
Spinal muscular atrophy, distal, autosomal recessive 1 (with respiratory distress)	12.5	AR	DSMA1 604320	11q13.3	***IGHMBP2*** *600502*	Immunoglobulin Mu-binding protein 2	Grohmann et al (1999, 2001)	allelic to CMT2S (group 14)
Spinal muscular atrophy, distal autosomal recessive 2	12.6	AR	DSMA2 605726	9p13.3	***SIGMAR1*** *601978*	Sigma non-opioid intracellular receptor 1	Christodoulou et al (2000) Li et al (2015)	allelic to ALS16 (group 12)
Spinal muscular atrophy, distal autosomal recessive 3	12.7	AR	DSMA3 607088	11q13	***?***		Viollet et al (2004)	
Spinal muscular atrophy, distal autosomal recessive 4	12.8	AR	DSMA4 611067	1p36.31	***PLEKHG5*** *611101*	Pleckstrin homology domain and RhoGEF domai-containing protein G5	Maystadt et al (2006, 2007)	allelic to CMTRIC (group 14)
Spinal muscular atrophy, distal, autosomal recessive, 5	12.9	AR	DSMA5 614881	2q35	***DNAJB2*** *604139*	DnaJ/Hsp40 homolog, subfamily B, member 2	Blumen et al (2012)	
Spinal muscular atrophy with congenital bone fractures 1	12.10	AR	SMABF1 616866	15q22.31	***TRIP4*** *604501*	Thyroid hormone receptor interactor 4	Knierim et al (2016)	allelic to MDCDC (group 2)
Spinal muscular atrophy with congenital bone fractures 2	12.11	AR	SMABF2 616867	10q22.1	***ASCC1*** *614215*	Activating signal cointegrator 1 complex subunit 1	Knierim et al (2016)	
Spinal muscular atrophy with progressive myoclonic epilepsy	12.12	AR	SMAPME 159950	8p22	***ASAH1*** *613468*	N-acylsphingosine amidohydrolase 1	Zhou et al (2012)	allelic to FRBRL (#228000)
Spinal muscular atrophy and cerebellar hypoplasia	12.13	AR	PCH1C 616081	13q13.3	***EXOSC8*** *606019*	Exosome component 8	Boczonadi et al (2014)	
Distal spinal muscular atrophy, dominant								
Neuronopathy, distal hereditary motor, type I	12.14	AD	HMN1 182960	7q34-q36	***?***		Gopinath et al (2007)	
Neuronopathy, distal hereditary motor, type IIA	12.15	AD	HMN2A 158590	12q24.23	***HSPB8*** *608014*	Heat-shock 22-kD protein 8	Timmerman et al (1992) Irobi et al (2004)	allelic to CMT2L (group 14)
Neuronopathy, distal hereditary motor, type IIB	12.16	AD	HMN2B 608634	7q11.23	***HSPB1*** *602195*	Heat-shock 27-kD protein 1	Evgrafov et al (2004)	allelic to CMT2F (group 14)
Neuronopathy, distal hereditary motor, type IIC	12.17	AD	HMN2C 613376	5q11.2	***HSPB3*** *604624*	Heat shock 27-kD protein 3	Kolb et al. (2010)	

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Neuronopathy, distal hereditary motor, type IID	12.18	AD	HMN2D 615575	5q32	***FBXO38*** *608533*	F-box only protein 38	Sumner et al (2013)	
Distal spinal muscular atrophy, distal with upper limb predominance (type V)	12.19	AD	HMN5A 600794	7p14.3	***GARS*** *600287*	Glycyl-tRNA synthetase	Christodoulou et al (1995) Antonellis et al (2003)	allelic to CMT2D (group 14)
Distal spinal muscular atrophy type VA	12.20	AD	HMN5A 600794	11q12.3	***BSCL2*** *606158*	Seipin	Windpassinger et al (2004)	allelic to SPG17 (group 15)
Distal spinal muscular atrophy, type VB	12.21	AD	HMNS5B 614751	2p11.2	***REEP1*** *609139*	Receptor expression-enhancing protein 1	Beetz et al (2012)	allelic to SPG31 (group 15)
Dominant distal hereditary motor neuropathy	12.22	AD	dHMN	16q22.1	***AARS*** *601065*	Alanyl-tRNA synthetase	Zhao et al (2012)	allelic to CMT2N (group 14)
Neuronopathy, distal hereditary motor, type IX	12.23	AD	HMN9 61772	14q32.2	***WARS*** *191050*	Tryptophanyl-tRNA synthetase	Tsai et al (2017)	
Spinal muscular atrophy, distal, with vocal cord paralysis (Harper-Young)	12.24	AD	HMN7A 158580	2q12.3	***SLC5A7*** *608761*	Solute carrier family 5 (choline cotransporter), member 7	McEntagart et al. (2001) Barwick et al (2012)	allelic to CMS20 (group 11)
Distal hereditary motor neuronopathy type VIIB	12.25	AD	HMN7B 607641	2p13.1	***DCTN1*** *601143*	Dynactin 1	Puls et al (2003)	allelic to ALS related to DNCT1 (group 12)
Hereditary motor and sensory neuropathy V	12.26	AD	HMSN5 600361	4q34.3-q35.2	*?*		Muglia et al (2008)	
Spinal muscular atrophy, distal, congenital non progressive	12.27	AD	HMN8 600175	12q24.11	***TRPV4*** *605427*	Transient receptor potential cation channel, subfamily V, member 4	van der Vleuten et al (1998) Auer-Grumbach et al (2010) Deng et al (2010)	allelic to SPSMA (group 12), CMT2C (group 14)
Scapuloperoneal spinal muscular atrophy	12.28	AD	SPSMA 181405	12q.24	***TRPV4*** *605427*	Transient receptor potential cation channel, subfamily V, member 4	Isozumi et al (1996) Deng et al (2010)	allelic to HMN8 (group 12), CMT2C (group 14)
Spinal motor neuropathy	12.29			11q23.2	***RBM7*** *612413*	RNA-binding motif protein 7	Giunta et al (2016)	
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 1	12.30	AD	SMALED1 158600	14q32.31	***DYNC1H1*** *600112*	Dynein, cytoplasmic 1, heavy chain 1	Harms et al (2010, 2012)	allelic to CMT2O (group 14)
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant 2	12.31	AD	SMALED2 615290	9q22.31	***BICD2*** *609797*	Bicaudal D homolog 2 (Drosophila)	Neveling et al (2013) Oates et al (2013) Peeters et al (2013)	
Spinal muscular atrophy, late-onset, Finkel type	12.32	AD	SMAFK 182980	20q13.32	***VAPB*** *605704*	Vesicle-associated membrane protein-associated protein B	Nishimura et al (2004)	allelic to ALS8 (group 12)
Spinal muscular atrophy, Jokela type	12.33	AD	SMAJ 615048	22q11.23	***CHCHD10*** *615903*	Coiled-coil-helix-coiled-coil-helix domain containing 10 (**M**)	Muller et al (2014) Penttilä et al (2012, 2015)	allelic to FTDALS2 (group 12), mitochondrial myopathy (group 16)
Distal spinal muscular atrophy, X-linked								
Spinal and bulbar muscular atrophy, X-linked, 1 (Kennedy disease)	12.34	XR	SBMX1 313200	Xq12	***AR*** *313700*	Androgen receptor	Fishbeck et al. (1986) La Spada et al. (1991)	
Spinal muscular atrophy, distal, X-linked 2	12.35	XR	SMAX2 301830	Xp11.3	***UBA1*** *314370*	Ubiquitin-like modifyer-activating enzyme 1	Ramser et al (2013) Diamini et al (2013)	
Spinal muscular atrophy, distal, X-linked, 3	12.36	XR	SMAX3 300489	Xq21.1	***ATP7A*** *300011*	ATPase, Cu^{++} transporting, alpha polypeptide	Takata et al (2004) Kennerson et al (2010)	
Amyotrophic lateral sclerosis (ALS)								
Amyotrophic lateral sclerosis 1 (dominant)	12.37	AD	ALS1 105400	21q22.11	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Siddique et al. (1991, 1996) Rosen et al. (1993)	
Amyotrophic lateral sclerosis 1 (recessive)	12.38	AR	ALS1 105400	21q22.11	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Andersen et al (1995)	
Amyotrophic lateral sclerosis 2, juvenile	12.39	AR	ALS2 205100	2q33.1	***ALS2*** *606352*	Alsin	Hentati et al. (1994) Yang et al (2001) Hadano et al (2001)	allelic to IAHSP (group 15)
Amyotrophic lateral sclerosis 3	12.40	AR	ALS3 606640	18q21	*?*		Hand et al (2002)	
Amyotrophic lateral sclerosis 4, juvenile	12.41	AD	ALS4 602433	9q34.13	***SETX*** *608465*	Senataxin	Chance et al (1998) Chen et al (2004) Moreira et al (2004)	allelic to SCAR1 (group 13)
Amyotrophic lateral sclerosis 5, juvenile	12.42	AR	ALS5 602099	15q21.1	***SPG11*** *610844*	Spatacsin	Hentati et al (1998) Orlacchio et al (2010)	allelic to CMT2X (group 14), SPG11 (group 15)

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Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	12.43	AD	ALS6 608030	16p11.2	***FUS*** *137070*	Fused in sarcoma	Sapp et al (2003) Abalkhail et al (2003) Kwiatkowski et al (2009) Vance et al (2009)	
Amyotrophic lateral sclerosis 7	12.44	AD	ALS7 608031	20p13	*?*		Sapp et al (2003)	
Amyotrophic lateral sclerosis 8	12.45	AD	ALS8 608627	20q13.32	***VAPB*** *605704*	Vesicle-associated membrane protein-associated protein B and C	Nishimura et al (2004)	allelic to SMAFK (group 12)
Amyotrophic lateral sclerosis 9	12.46	AD	ALS9 611895	14q11.2	***ANG*** *105850*	Angiogenin	Greenway et al (2006) Wu et al (2007)	
Amyotrophic lateral sclerosis 10, with or without frontotemporal dementia	12.47	AD	ALS10 612069	1p36.22	***TARDBP*** *605078*	TAR DNA-binding protein	Sreedharan et al (2008)	
Amyotrophic lateral sclerosis 11	12.48	AD	ALS11 612577	6q21	***FIG4*** *609390*	FIG4, S. Cerevisiae, homolog of	Chow et al (2009)	allelic to CMT4J (group 14)
Amyotrophic lateral sclerosis 12	12.49	AD, AR	ALS12 613435	10p13	***OPTN*** *602432*	Optineurin	Maruyama et al (2010)	
Amyotrophic lateral sclerosis 13	12.50	AD	ALS13 183090	12q24.12	***ATXN2*** *601517*	Ataxin 2	Elden et al (2010) Daoud et al (2011) Van Damme et al (2011)	allelic to SCA2 (group 13)
Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia	12.51	AD	ALS14 613954	9p13.3	***VCP*** *601023*	Valosin-containing protein	Johnson et al (2011)	allelic to scapuloperoreal MD (group 1), IBMPFD (groups 4 and 5), CMT2Y (group 14)
Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	12.52	XD	ALS15 300857	Xp11.21	***UBQLN2*** *300264*	Ubiquilin 2	Deng et al (2011)	
Amyotrophic lateral sclerosis 16, juvenile	12.53	AR	ALS16 614373	9p13.3	***SIGMAR1*** *601978*	Sigma non-opioid intracellular receptor 1	Al-Saif et al (2011)	allelic to DSMA2 (group 12)
Amyotrophic lateral sclerosis 17	12.54	AD	ALS17 614696	3p11.2	***CHMP2B*** *609512*	Charged multivesicular body protein 2B	Parkinson et al (2006) Cox et al (20010)	
Amyotrophic lateral sclerosis 18	12.55	AD	ALS18 614808	17p13.2	***PFN1*** *176610*	Profilin 1	Wu et al (2012)	
Amyotrophic lateral sclerosis 19	12.56	AD	ALS19 615515	2q34	***ERBB4*** *600543*	V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 4	Takahashi et al (2013)	
Amyotrophic lateral sclerosis 20	12.57	AD	ALS20 615426	12q13.13	***HNRNPA1*** *164017*	Heterogeneous nuclear ribonucleoprotein A1	Kim et al (2013)	allelic to IBMPFD3 (group 3)
Amyotrophic lateral sclerosis 21	12.58	AD	ALS21 606070	5q31.2	***MATR3*** *164015*	Matrin 3	Johnson et al (2014)	allelic to VCPDM (group 4)
Amyotrophic lateral sclerosis 22, with or without frontotemporal dementia	12.59	AD	ALS22 616208	2q35	***TUBA4A*** *191110*	Tubulin, alpha-4A	Smith et al (2014)	
Amyotrophic lateral sclerosis 23	12.60	AD	ALS23 617839	10q22.3	***ANXA11*** *602572*	Annexin A11	Smith et al (2017)	
Amyotrophic lateral sclerosis 24, suseptibility to	12.61		ALS24 617892	4q33	***NEK1*** *604588*	Never in mitosis gene A-related kinase 1	Brenner et al (2016) Kenna et al (2016)	
Amyotrophic lateral sclerosis related to NEFH, susceptibility to	12.62	AD, AR	ALS1 105400	22q12.2	***NEFH*** *162230*	Neurofilament, heavy polypeptide	Al-Chalabie et al (1999)	allelic to CMT2CC (group 14)
Amyotrophic lateral sclerosis related to peripherin, susceptibility to	12.63	AD	ALS1 105400	12q13.12	***PRPH*** *170710*	Peripherin	Gros-Louis et al (2004) Leung et al (2004)	
Amyotrophic lateral sclerosis related to dynactin 1, susceptibility to	12.64	AD	ALS1 105400	2p13.1	***DCTN1*** *601143*	Dynactin 1	Munch et al (2005)	allelic to HMN7B (group 12)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.65	AD	FTDALS1 105550	9p21.2	***C9orf72*** *614260*	Chromosome 9 open reading frame 72	Morita et al (2006) DeJesus-Hernandez (2011)	
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.66	AD	FTDALS2 615911	22q11.23	***CHCHD10*** *615903*	Coiled-coil-helix-coiled-coil-helix domain containing 10 (**M**)	Bannwarth et al (2014)	allelic to SMAJ (group 12), mitochondrial myopathy (group 16)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.67	AD	FTDALS3 66437	5q35.3	***SQSTM1*** *601530*	Sequestosome 1	Fecto et al (2011)	allelic to DMRV (group 4)

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Others								
Lethal Congenital Contracture Syndrome 1	12.68	AR	LCCS1 253310	9q34.11	***GLE1*** *603371*	GLE1, RNA export mediator	Makela-Bengs et al (1998) Nousiainen et al. (2008)	
Lethal Congenital Contracture Syndrome 2	12.69	AR	LCCS2 607598	12q13.2	***ERBB3*** *190151*	V-ERB-B2 avian erythroblastic leukemia viral oncogene homolog 3	Narkis et al (2007)	
Lethal Congenital Contracture Syndrome 3	12.70	AR	LCCS3 611359	19p13.3	***PIP5K1C*** *606102*	Phosphatidylinositol-4-phosphate 5-kinase, type I, gamma	Narkis et al (2007)	
Spinal muscular atrophy with pontocerebellar hypoplasia, type 1b	12.71	AR	PCH1B 614678	9p13.2	***EXOSC3*** *606489*	Exosome component 3	Rudnik-Schöneborn et al (2013)	
Spinal muscular atrophy with or without pontocerebellar hypoplasia	12.72	AR	PCH1 607596	14q32.2	***VRK1*** *602168*	Vaccinia related kinase 1	Renbaum et al (2009) Stoll et al (2016)	
Pontocerebellar hypoplasia with spinal muscular atrophy	12.73	AR	HSMN6B 616505	5q22.1	***SLC25A46*** *610826*	Solute Carrier Family 25, Member 46	van Dijk et al (2017)	
Brown-Vialetto-van Laere syndrome 1	12.74	AR	BVVLS1 211530	20p13	***SLC52A3*** *613350*	Solute carrier family 52 (riboflavin transporter) member 3	Green et al (2010) Bosch et al (2011) Johnson et al (2010)	
Brown-Vialetto-van Laere syndrome 2	12.75	AR	BVVLS2 614707	8q24.3	***SLC52A2*** *607882*	Solute carrier family 52 (riboflavin transporter) member 2	Green et al (2010) Bosch et al (2011) Johnson et al (2012)	
Late onset spinal muscular atrophy related to *HEXB*	12.76	AR		5q13.3	***HEXB*** *606873*	Hexosamin-idase B	Rattay et al (2013)	allelic to Sandhoff disease, adult type (OMIM #268800)
Spinal muscular atrophy, related to PRUNE1	12.77	AR		1q21.3	***PRUNE1*** *617413*	Prune exopolyphos-phatase 1	Iacomino et al (2017)	
GROUP 13. HEREDITARY ATAXIAS. See online version of the gene table http://www.musclegenetable.fr								
GROUP 14. HEREDITARY MOTOR SENSORY NEUROPATHIES (HMSN)								
A. Charcot-Marie-Tooth neuropathy, type 1 (demyelinating)								
Autosomal dominant (AD-CMT1)								
Charcot-Marie-Tooth disease, type 1A	14.1	AD	CMT1A 118220	17p12	***PMP22*** *601097*	Peripheral myelin protein 22	Vance et al (1989) Patel et al (1992) Matsunami et al (1992) Timmerman et al (1990, 1992) Valentijn et al (1992) Roa et al (1993)	allelic to CMT1E, HNPP and DSS (group 14)
Charcot-Marie-Tooth disease, type 1B	14.2	AD	CMT1B 118200	1q23.3	***MPZ*** *159440*	Myelin protein zero	Bird et al (1982) Guiloff et al (1982) Hayasaka et al (1993) Kulkens et al (1993)	allelic to CMTDID, CMT2I, CMT2J, DSS and CHN (group14)
Charcot-Marie-Tooth disease, type 1C	14.3	AD	CMT1C 601098	16p13.13	***LITAF*** *603795*	Lipopoly-saccharide-induced TNF-alpha factor	Street et al (2002, 2003)	
Charcot-Marie-Tooth disease, type 1D	14.4	AD	CMT1D 607678	10q21.3	***EGR2*** *129010*	Early growth response 2 (Krox-20 homolog)	Warner et al (1998) Street et al (2003)	allelic to CHN and DSS (group 14)
Charcot-Marie-Tooth disease, type 1E, with deafness	14.5	AD	CMT1E 118300	17p12	***PMP22*** *601097*	Peripheral myelin protein 22	Kovach et al (1999) Boerkoel et al (2002)	allelic to CMT1A, HNPP and DSS (group14),
Hereditary neuropathy with liability to pressure palsies	14.6	AD	HNPP 162500	17p12	***PMP22*** *601097*	Peripheral myelin protein P22	Chance et al (1993) Nicholson et al (1994) Mariman et al (1994)	allelic to CMT1A, CMT1E and DSS (group 14)
Charcot-Marie-Tooth disease, type 1F	14.7	AD	CMT1F 607734	8p21.2	***NEFL*** *162280*	Neurofilament, light polypeptide 68kDa	Jordanova et al (2003)	allelic to CMTDIG and CMT2E (group 14)
CMT with congenital vertical talus	14.8	AD	CVT 192950	2q31.1	***HOXD10*** *142984*	Homeobox D10	Shrimpton et al (2004)	
Slowed nerve conduction velocity	14.9	AD	SNCV 608236	8p23.3	***ARHGEF10*** *608136*	Rho guanine-nucleotide exchange factor-10	De Jonghe et al (1999) Verhoeven et al (2003)	
Neuropathy, hereditary, with or without age-related macular degeneration	14.10	AD	HNARMD 608895	14q32.12	***FBLN5*** *604580*	Fibulin 5	Auer-Grumbach et al (2011)	

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CMT1 related to *PMP2*	14.11	AD		8q21.13	***PMP2*** *170715*	Peripheral myelin protein 2	Hong et al (2016)	
Dominant intermediate (CMTDI)								
Charcot-Marie-Tooth disease, dominant intermediate A	14.12	AD	CMTDIA 606483	10q24.1-q25.1	***?***		Verhoeven et al (2001)	
Charcot-Marie-Tooth disease, dominant intermediate B	14.13	AD	CMTDIB 606482	19p13.2	***DNM2*** *602378*	Dynamin 2	Zuchner et al (2005)	allelic to CNM1 (group 3) and CMD (group 2)
Charcot-Marie-Tooth disease, dominant intermediate C	14.14	AD	CMTDIC 608323	1p35.1	***YARS*** *603623*	Tyrosyl-tRNA synthetase	Jordanova et al (2003, 2006)	
Charcot-Marie-Tooth disease, dominant intermediate D	14.15	AD	CMTDID 607791	1q23.3	***MPZ*** *159440*	Myelin protein zero	Mastaglia et al (1999)	allelic to CMT1B, CMTDID, CMT2I, CMT2J, DSS and CHN (group14)
Charcot-Marie-Tooth disease, dominant intermediate E	14.16	AD	CMTDIE 614455	14q32.33	***INF2*** *610982*	Inverted formin 2	Boyer et al (2011)	
Charcot-Marie-Tooth disease, dominant intermediate F	14.17	AD	CMTDIF 615185	3q28.33	***GNB4*** *610863*	Guanine nucleotide-binding protein, beta-4	Soong et al (2013)	
Charcot-Marie-Tooth disease, dominant intermediate G	14.18	AD	CMTDIG 617882	8p21.2	***NEFL*** *162280*	Neurofilament, light polypeptide 68kDa	Berciano et al (2015)	allelic to CMT1F and CMT2E (group 14)
Autosomal recessive (AR-CMT1 or CMT4)								
Charcot-Marie-Tooth disease, type 4A	14.19	AR	CMT4A 214400	8q21.11	***GDAP1*** *606598*	Ganglioside-induced differentiation associated protein1	Ben Othmane et al (1993) Baxter et al (2002) Cuesta et al (2002) Nelis et al (2002)	allelic to CMT2K and CMTRIA (group 14)
Charcot-Marie-Tooth disease, type 4B1	14.20	AR	CMT4B1 601382	11q21	***MTMR2*** *603557*	Myotubularin-related protein-2	Bolino et al (1996, 2000) Previtali et al (2003)	
Charcot-Marie-Tooth disease, type 4B2	14.21	AR	CMT4B2 604563	11p15.4	***SBF2*** *607697*	SET-binding factor 2	Azzedine et al (2003) Senderek et al (2004)	
Charcot-Marie-Tooth disease, type 4B3	14.22	AR	CMT4B3 615284	22q13.33	***SBF*** *603560*	SET-binding factor 1	Nakhro et al (2013)	
Charcot-Marie-Tooth disease, type 4C	14.23	AR	CMT4C 601596	5q32	***SH3TC2*** *608206*	SH3 domain and tetratricopeptide repeats domain 2	LeGuern et al (1996) Senderek et al (2003)	
Charcot-Marie-Tooth disease, type 4D	14.24	AR	CMT4D 601455	8q24.22	***NDRG1*** *605262*	N-myc downstream regulated gene 1	Kalaydjieva et al (1996, 2000) Hunter et al (2003)	
Neuropathy, congenital hypomyelinating, 1	14.25	AR	CHN 605253	10q21.3	***EGR2*** *129010*	Early growth response 2	Warner et al (1998)	allelic to CMT1D and DSS (group 14)
Neuropathy, congenital hypomyelinating, 1	14.26	AR	CHN 605253	1q23.3	***MPZ*** *159440*	Myelin protein zero	Warner et al. (1996)	allelic to CMT1B, CMTDID, CMT2I, CMT2J, DSS (group 14)
Charcot-Marie-Tooth disease, type 4F	14.27	AR	CMT4F 614895	19q13.2	***PRX*** *605725*	Periaxin	Delague et al (2000) Guilbot et al (2001)	allelic to DSS (group14)
Neuropathy, hereditary motor and sensory, Russe type	14.28	AR	HMSNR 605285	10q22.1	***HK1*** *142600*	Hexokinase 1	Rogers et al (2000) Thomas et al (2001) Hantke et al (2009) Sevilla et al (2013)	
Charcot-Marie-Tooth disease, type 4H	14.29	AR	CMT4H 609311	12p11.21	***FGD4*** *611104*	Fyve, RhoGEF and Phdomain-containing protein 4 (Frabin)	De Sandre-Giovannoli et al (2005) Delague et al (2007) Stendel et al (2007)	
Charcot-Marie-Tooth disease, type 4J	14.30	AR	CMT4J 611228	6q21	***FIG4*** *609390*	FIG4, S. Cerevisiae, homolog of	Chow et al (2007)	allelic to ALS11 (group 12)
Charcot-Marie-Tooth disease, type 4K	14.31	AR	CMT4K 616684	9q34.2	***SURF1*** *185620*	Surfeit 1 (**M**)	Echaniz-Laguna et al (2013)	
X-linked CMT1								
Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	14.32	XD	CMTX1 302800	Xq13.1	***GJB1*** *304040*	Gap junction protein, beta 1	Bergoffen et al (1993) Bone et al (1995)	allelic to DSS (group 14)
Charcot-Marie-Tooth neuropathy, X-linked recessive, 2	14.33	XR	CMTX2 302801	Xp22.2	***?***		Ionasecu et al (1992)	
Charcot-Marie-Tooth neuropathy, X-linked recessive, 3	14.34	XR	CMTX3 302802	Xq26	***78kb Chro8 insertion***	78kb inter-chromosomal insertion (from chro 8q24.3)	Ionasecu et al (1992) Huttner et al (2006) Brewer et al (2016)	
Charcot-Marie-Tooth disease, X-linked 4 (Cowchock syndrome)	14.35	XR	COWCK 310490	Xq26.1	***AIFM1*** *300169*	Apoptosis-inducing factor, mitochondria-associated, 1 (**M**)	Priest et al (1995) Rinaldi et al (2012)	
Charcot-Marie-Tooth disease, X-linked recessive, 5	14.36	XR	CMTX5 311070	Xq22.3	***PRPS1*** *311850*	Phosphoribosyl pyrophosphate synthetase 1	Kim at al (2007)	

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Charcot-Marie-Tooth disease, X-linked dominant, 6	14.37	XD	CMTX6 300905	Xp22.11	***PDK3*** *300906*	Pyruvate dehydrogenase kinase, isoenzyme 3	Kennerson et al (2013)	
Déjerine-Sottas syndrome (DSS or CMT3)								
Déjerine-Sottas hypertrophic neuropathy, dominant	14.38	AD	DSS 145900	17p12	***PMP22*** *601097*	Peripheral myelin protein 22	Roa et al (1993)	allelic to CMT1A, CMT1E and HNPP (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.39	AD	DSS 145900	1q23.3	***MPZ*** *159440*	Myelin protein zero	Hayasaka et al (1993)	allelic to CMT1B, CMTD2I, CMT2J, CMT2I and CHN (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.40	AD (digenic)	DSS 145900	10q21.3 and Xq13	***EGR2*** *129010* and ***GJB1*** *304040*	Early growth response 2 *and* Gap junction protein, beta 1	Chung et al (2005)	allelic to CMTX1 (group 14)
Déjerine-Sottas hypertrophic neuropathy, recessive	14.41	AR	DSS 145900	19q13.2	***PRX*** *605725*	Periaxin	Delague et al (2000) Boerkoel et al (2001)	allelic to CMT4F (group 14)
B. Charcot-Marie-Tooth neuropathy, type 2 (axonal) = CMT2 Autosomal dominant CMT2								
Charcot-Marie-Tooth disease, type 2A1	14.42	AD	CMT2A1 118210	1p36.22	***KIF1B*** *605995*	Kinesin family member 1B	Zhao et al (2001)	
Charcot-Marie-Tooth disease, axonal, type 2A2A	14.43	AD	CMT2A2A 609260	1p36.22	***MFN2*** *608507*	Mitofusin 2	Ben Othmane et al (1993) Züchner et al (2004)	allelic to CMT2A2B and HMSN6A (group14)
Charcot-Marie-Tooth disease, type 2B	14.44	AD	CMT2B 600882	3q21.3	***RAB7*** *602298*	RAS-asociated proteinRAB7	Kwon et al (1995) Pericak-Vance et al (1997) Kok et al (2003)	
Charcot-Marie-Tooth disease, type 2C	14.45	AD	HMSN2C 606071	12q24.11	***TRPV4*** *605427*	Transient receptor potential cation channel, subfamily V, member 4	Klein et al (2003) McEntagart et al (2005) Auer-Grumbach et al (2010) Deng et al (2010) Landoure et al (2010)	allelic to distal SMA and SPSMA (group 12)
Charcot-Marie-Tooth disease, type 2D	14.46	AD	CMT2D 601472	7p14.3	***GARS*** *600287*	Glycyl-tRNA synthetase	Ionasescu et al (1996) Antonellis et al (2003)	allelic to HMN5A (group12)
Charcot-Marie-Tooth disease, type 2E	14.47	AD	CMT2E 607684	8p21.2	***NEFL*** *162280*	Neurofilament, light polypeptide	Birouk et al (2003) Claramunt et al (2005)	allelic to CMT1F, CMTDIG (group 14)
Charcot-Marie-Tooth disease, type 2F	14.48	AD	CMT2F 606595	7q11.23	***HSPB1*** *602195*	Heat-shock 27-kD protein 1	Ismailov et al (2001) Evgrafov et al (2004)	allelic to HMN2B (group 14)
Charcot-Marie-Tooth disease, type 2H	14.49	AD	CMT2H 607731	8q13-q23	*?*	?	Barhoumi et al (2001)	maybe allelic to CMT4A (group14)
Charcot-Marie-Tooth disease, type 2I	14.50	AD	CMT2I 607677	1q23.3	***MPZ*** *159440*	Myelin protein zero	Auer-Grumbach et al (2003)	allelic to CMT1B, CMTDID, CMT2J, DSS, CHN (group 14)
Charcot-Marie-Tooth disease, type 2J	14.51	AD	CMT2J 607736	1q23.3	***MPZ*** *159440*	Myelin protein zero	De Jonghe et al (1999) Chapon et al (1999)	allelic to CMT1B, CMTDID, CMT2I, DSS, CHN (group 14)
Charcot-Marie-Tooth disease, type 2K	14.52	AD, AR	CMT2K 607831	8q21.11	***GDAP1*** *606598*	Ganglioside-induced differentiation-associated protein 1	Nelis et al (2002) Birouk et al (2003) Claramunt et al (2005)	allelic to CMT4A and CMTRIA (group 14)
Charcot-Marie-Tooth disease, type 2L	14.53	AD	CMT2L 608673	12q24.23	***HSPB8*** *608014*	Heat-shock 22-kD protein 8	Tang et al (2004, 2005)	allelic to HMN2A (group 12)
Charcot-Marie-Tooth disease, type 2N	14.54	AD	CMT2N 613287	16q22.1	***AARS*** *601065*	Alanyl-tRNA synthetase	Latour et al (2010)	
Charcot-Marie-Tooth disease, type 2O	14.55	AD	CMT2O 614228	14q32.31	***DYNC1H1*** *600112*	Dynein, cytoplasmic 1, heavy chain 1	Weedon et al (2011)	allelic to SMALED1 (group 12)
Charcot-Marie-Tooth disease, type 2P	14.56	AD	CMT2P 614436	9q33.3-q34.1	***LRSAM1*** *610933*	Leucine rich repeat and sterile alpha motif containing 1	Nelis et al (2004) Guernsey et al (2010) Weterman et al (2012) Nicolaou et al (2012) Peeters et al (2016)	
Charcot-Marie-Tooth disease, type 2Q	14.57	AD	CMT2Q 615025	10p14	***DHTKD1*** *614984*	Dehydrogenase E1 and transketolase domain containing 1	Xu et al (2012)	
Charcot-Marie-Tooth disease, type 2U	14.58	AD	CMT2U 616280	12q13.3	***MARS*** *156560*	Methionyl-tRNA synthetase	Gonzalez et al (2013)	
Charcot-Marie-Tooth disease, type 2V	14.59	AD	CMT2V 616491	17q21.2	***NAGLU*** *609701*	N-acetyl-alpha-glucosaminidase	Tetreault et al (2015)	
Charcot-Marie-Tooth disease, type 2W	14.60	AD	CMT2W 616625	5q31.3	***HARS*** *142810*	Histidyl-tRNA synthetase	Vester et al (2013) Safka-Brozkova et al (2015)	
Charcot-Marie-Tooth disease, type 2Y	14.61	AD	CMT2Y 616687	9p13.3	***VCP*** *601023*	Valosin-containing protein	Gonsalez et al (2014)	allelic to scapuloperoreal MD (group 1), IBMPFD (groups 4 and 5), ALS14 (group 12)

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Charcot-Marie-Tooth disease, type 2Z	14.62	AD	CMT2Z 616688	22q12.2	***MORC2*** *616661*	MORC family CW-type zinc finger 2	Albulym et al (2015) Sevilla et al (2016)	
Hereditary motor and sensory neuropathy, Okinawa type	14.63	AD	HMSNO 604484	3q12.2	***TFG*** *602498*	TRK-fused gene	Takeshima et al (1997, 1999) Maeda et al (2007) Ishiura et al (2012)	allelic to SPG57 (group 15)
CMT2 related to *KIF5A*	14.64	AD		12q13.3	***KIF5A*** *602821*	Kinesin family member 5A	Liu et al (2014)	allelic to SPG10 (group 15)
Early onset axonal neuropathy with sensory ataxia	14.65	AD	CMT2	1q13.5	***DGAT2*** 606983	Diacylglycerol O-acyltransferase 2	Hong et al (2016)	
Charcot-Marie-Tooth disease, axonal, type 2CC	14.66	AD	CMT2CC 616924	22q12.2	***NEFH*** *162230*	Neurofilament Protein, Heavy Polypeptide	Rebelo et al (2016)	allelic to ALS (group 12)
Charcot-Marie-Tooth disease, axonal, related to BAG3	14.67	AD		10q26.11	***BAG3*** *603883*	BCL2-associated athanogene 3	Noury et al (2017)	allelic to MFM6 (group 5) and to CMH1HH (group 10)
Charcot-Marie-Tooth disease, axonal, type 2DD	14.68	AD	CMT2DD 618036	1p13.1	***ATP1A1*** *182310*	ATPase, Na^+/K^+ transporting, alpha-1 polypeptide	Lassuthova et al (2018)	
Autosomal recessive CMT2								
Charcot-Marie-Tooth disease, axonal, type 2A2B	14.69	AR	CMT2A2B 617087	1p36.22	***MFN2*** *608507*	Mitofusin 2	Polke et al (2011)	allelic to CMT2A2A and HMSN6A (group14)
Charcot-Marie-Tooth disease, axonal, type 2B1	14.70	AR	CMT2B1 605588	1q22	***LMNA*** *150330*	Lamin A/C	Bouhouch et al (1999) De Sandre et al (2002) Worman and Bonne (2007)	allelic to EDMD2, EDMD3, LGMD1B (group1), MDCL (group 2), CMD1A (group 10) [+ several other phenotypes not in this table: FPLD2 #151660, HGPS #176670, restrictive dermopathy #275210, MADA #248370]
Charcot-Marie-Tooth disease, axonal, type 2B2	14.71	AR	CMT2B2 605589	19q13.33	***MED25*** *610197*	Mediator complex subunit 25	Leal et al (2001, 2009)	
Charcot-Marie-Tooth disease, axonal, related to *DNAJB2*	14.72	AR	DSM5 614881	2q35	***DNAJB2*** *604139*	DnaJ/Hsp40 homolog, subfamily B, member 2	Gess et al (2014)	allelic to SMA (group 12)
Neuromyotonia and axonal neuropathy, autosomal recessive	14.73	AR	NMAN 137200	5q23.3	***HINT1*** *601314*	Histidine triad nucleotide binding protein 1	Zimon et al (2012)	
Charcot-Marie-Tooth disease, recessive intermediate, A	14.74	AR	CMTRIA 608340	8q21.11	***GDAP1*** *606598*	Ganglioside-induced differentiation-associated protein 1	Nelis et al (2002)	allelic to CMT4A and CMT2K (group 14)
Charcot-Marie-Tooth disease, recessive intermediate, B	14.75	AR	CMTRIB 613641	16q23.1	***KARS*** *601421*	Lysyl-tRNA synthetase	McLaughlin et al (2010)	
Charcot-Marie-Tooth disease, recessive intermediate, C	14.76	AR	CMTRIC 615376	1p36.31	***PLEKHG5*** *611101*	Pleckstrin homology domain- and RhoGEF domain-containing, family G5	Azzedine et al (2013) Kim et al (2013)	allelic to DSMA4 (group 12)
Charcot-Marie-Tooth disease, recessive intermediate, D	14.77	AR	CMTRID 616039	12q24.31	***COX6A1*** *602072*	Cytochrome c oxidase subunit 6a1 (**M**)	Tamiya et al (2014)	
Charcot-Marie-Tooth disease, type 2R	14.78	AR	CMT2R 615490	4q31.3	***TRIM2*** *614141*	Tripartite motif-containing protein 2	Ylikallio et al. (2013) Pehlivan et al (2015)	
Charcot-Marie-Tooth disease, type 2S	14.79	AR	CMT2S 616155	11q13.3	***IGHMBP2*** *600502*	Immunoglobulin mu- binding protein 2	Cottenie et al (2014) Schottmann et al (2015)	allelic to DSMA1 (group 12)
Charcot-Marie-Tooth disease, type 2T	14.80	AR	CMT2T 617017	3q25.2	***MME*** *120520*	Membrane metal-loendopeptidase	Higuchi et al (2016)	allelic to SCA43 (group 15)
Charcot-Marie-Tooth disease, type 2X	14.81	AR	CMT2X 616668	15q21.1	***SPG11*** *610844*	SPG11 gene (Spatacsin)	Montecchiani et al (2015)	allelic to ALS5 (group 12), SPG11 (group 15)
Early-onset axonal Charcot-Marie-Tooth with ataxia	14.82	AR	AOA4 616267	19q13.33	***PNKP*** 605610	Polynucleotide kinase 3′-phosphatase	Pedroso et al (2015)	
Charcot-Marie-Tooth disease, axonal	14.83	AR		10q22.1	***SGPL1*** *603729*	Sphingosine-1 phosphate lyase 1	Atkinson et al (2017)	allelic to Nephrotic Syndrome 14 (# 617575)
Charcot-Marie-Tooth disease, axonal; related to *SCO2*	14.84	AR		22q13.33	***SCO2*** *604272*	Cytochrome c oxidase assembly protein	Rebelo et al (2018)	allelic to CEMCOX1 (group 10A)

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C. CMT Distal = Distal hereditary motor neuropathies (dHMN) = spinal CMT or distal spinal muscular atrophy (DSMA) See under MOTOR NEURONE DISEASES (Group 12)								
D. Other Hereditary sensory and autonomic neuropathy (HSAN)								
Hereditary sensory and autonomic neuropathy, type IA	14.85	AD	HSAN1A 162400	9q22.31	***SPTLC1*** *605712*	Serine palmitoyl transferase long-chain base subunit 1	Nicholson et al (1996) Bejaoui et al (2001) Dawkins et al (2001)	
Hereditary sensory and autonomic neuropathy, type IB with cough and gastroesophageal reflux	14.86	AD	HSAN1B 608088	3p24-p22	***?***		Kok et al (2004)	
Hereditary sensory and autonomic neuropathy, type IC	14.87	AD	HSAN1C 613640	14q24.3	***SPTLC2*** *605713*	Serine palmitoyl transferase long-chain base subunit 2	Rotthier et al (2010)	
Hereditary sensory and autonomic neuropathy, type IIA	14.88	AR	HSAN2A 201300	12p.13.33	***WNK1*** *605232*	Protein kinase, lysine deficient 1	Lafreniere et al (2004) Shekarabi et al (2008)	
Hereditary sensory and autonomic neuropath, type IIB	14.89	AR	HSAN2B 613115	5p15.1	***RETREG1*** 613114	Family with sequence similarity 134 member B	Kurth et al (2009)	
Hereditary sensory and autonomic neuropathy, type IID	14.90	AR	CIP 24300	2q24.3	***SCN9A*** 603415	Sodium channel, voltage-gated alpha subunit	Yuan et al (2013)	
Hereditary sensory and autonomic neuropathy type III	14.91	AR	HSAN3 223900	9q31.3	***ELP1*** *603722*	Inhibitor of kappa light polypeptide gene enhanccer in B cells, kinase complex associated protein (IKBKAP)	Blumenfeld et al (1993) Anderson et al (2001) Slaugenhaupt et al (2001)	allelic to familial Dysautonomia (group 16)
Hereditary sensory and autonomic neuropathy type IV	14.92	AR	CIPA 256800	1q23.1	***NTRK1*** 191315	Neurotrophic tyrosine kinase, receptor, type 1	Indo et al (1996)	
Hereditary sensory and autonomic neuropathy type V	14.93	AR	HSAN5	1p13.1	***NGFB*** *162030*	Nerve growth factor (beta polypeptide)	Einarsdottir et al (2004)	
Hereditary sensory and autonomic neuropathy type VI	14.94	AR	HSAN6 614653	6p12.1	***DST*** 113810	Dystonin	Edvardson et al (2012)	
Neuropathy, hereditary sensory and autonomic, type VII	14.95	AD	HSAN7 615548	3p22.2	***SCN11A*** 604385	Sodium channel, voltage-gated alpha subunit	Leipold et al (2013)	
Hereditary sensory and autonomic neuropathy type VIII	14.96	AR	HSAN8 616488	9q34.12	***PRDM12*** *616458*	PR/SET domain 12 (positive regulatory domain zinc finger protein 12)	Chen et al (2015)	
Hereditary sensory neuropathy, type ID	14.97	AD	HSN1D 613708	14q22.1	***ATL1*** *606439*	Atlastin GTPase 1	Guelly et al (2011)	allelic to SPG3A (group 15)
Hereditary sensory neuropathy, type IE	14.98	AD	HSN1E 614116	19p13.2	***DNMT1*** *126375*	DNA methyltransferase 1	Klein et al (2011)	
Neuropathy, hereditary sensory, type IF	14.99	AD	HSN1F 615632	11q13.1	***ATL3*** 609369	Atlastin GTPase 3	Kornak et al (2014)	
Hereditary sensory neuropathy, type IIC	14.100	AR	HSN2C 614213	2q37.3	***KIF1A*** *601255*	Kinesin family member 1A	Riviere et al (2011)	allelic to SPG30 (group 15)
Ataxia, posterior column, with retinitis pigmentosa (PCARP)	14.101	AR	AXPC1 609033	1q32.3	***FLVCR1*** *609144*	Feline leukemia subgroup C receptor 1	Rajadhyaksha et al (2010)	
Absence of pain, Congenital	14.102	AR		22q11.21	***CLTCL1 601273***	Clathrin, heavy polypepdide-like 1	Nahorski et al (2015)	
Marsili syndrome (insensitivity to pain, congenital, AD)	14.103	AD	MARSIS 147430	14q11.2	***ZFHX2*** *617828*	Zinc finger homeobox 2	Habib et al (2018)	
E. Other complex neuropathy syndromes								
Peripheral neuropathy and agenesis of the corpus callosum (Charlevoix disease)	14.104	AR	ACCPN 218000	15q14	***SLC12A6 (KCC3)*** *604878*	Solute carrier family 12 (potassium/ chloride transporter), member 6	Casaubon et al (1996) Howard et al (2002)	
Peripheral neuropathy and deafness, autosomal dominant	14.105	AD		1p34.3	***GJB3*** *603324*	Gap junction protein, beta 3	Lopez-Bigas et al (2001)	
Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy)	14.106	AD	HNA 162100	17q25.3	***SEPT9*** *604061*	Septin 9	Pellegrino et al (1996) Kuhlenbaumer et al (2005)	
Giant axonal neuropathy-1	14.107	AR	GAN1 256850	16q23.2	***GAN1*** *605379*	Gigaxonin	Ben Hamida et al (1997) Bomont et al (2000)	

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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)
Giant axonal neuropathy-2	14.108	AD	GAN2 610100	1q23.2	***DCAF8*** *615820*	DDB1- and CUL4-associated factor 8	Klein et al (2014)	
Congenital cataracts, facial dysmorphism and neuropathy	14.109	AR	CCFDN 604168	18p23	***CTDP1*** *604927*	C-terminal domain of RNA polymerase II subunit A, phosphatase of, subunit 1	Varon et al (2003)	
Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis	14.110	AR		14q32.2	***VRK1*** *602168*	Vaccinia related kinase 1	Gonzaga-Jauregui et al (2013)	
Neuropathy, hereditary sensory, with spastic paraplegia	14.111	AR	256840	5p15.2	***CCT5*** *610150*	Chaperonin containing T-complex polypeptide1, subunit 5	Bouhouche et al (2006)	
GROUP 15. HEREDITARY PARAPLEGIAS. ***See online version of the gene table*** http://www.musclegenetable.fr								
GROUP 16. OTHER NEUROMUSCULAR DISORDERS								
Torsion dystonia, early onset	16.1	AD	DYT1 128100	9q34.11	***TOR1A*** *605204*	Torsin 1A	Ozelius et al (1997) Ikeuchi et al (1999)	
Myoclonus-dystonia syndrome	16.2	AD	DYT11 159900	7q21.3	***SGCE*** *604149*	Sarcoglycan, Epsilon	Klein et al (2000) Zimprich et al (2001) Tezenas du Montcel et al (2006)	
Familial dysautonomia (Riley-Day syndrome)	16.3	AR	HSAN3 223900	9q31.3	***ELP*** *603722*	Inhibitor of kappa Light polypeptide gene enhaner in B cells, kinase complex associated protein	Blumenfeld et al (1993) Anderson et al (2001) Slaugenhaupt et al (2001)	allelic to HSNA3 (group 14)
Familial amyloid neuropathy	16.4	AD	105210	18q12.1	***TTR*** *176300*	Transthyretin	Costa et al. (1978) Tawara et al. (1983) Saraiva et al (1995)	
Fibrosis of extraocular muscles, congenital, 1	16.5	AD	CFEOM1 CFEOM3B 135700	12q12	***KIF21A*** *608283*	Kinesin family member 21A	Engle et al. (1994) Yamada et al (2003) Tiab et al (2004)	
Fibrosis of extraocular muscles, congenital, 2	16.6	AD	CFEOM2 602078	11q13.4	***PHOX2A*** *602753*	Aristaless homeobox, drosophila, homolg of, (ARIX)	Wang et al (1998) Nakano et al (2001)	
Fibrosis of extraocular muscles, congenital, 3A	16.7	AD	CFEOM3A 600638	16q24	***TUBB3*** *602661*	Tubulin, beta-3	Doherty et al (1999) Tischfield et al (2010)	
Fibrosis of extraocular muscles, congenital, 5	16.8	AR	CFEOM5 66219	4q25	***COL25A1*** *610004*	Collagen, type XXV, alpha-1	Shinwari et al (2015)	
Arthrogryposis, distal, type 1A	16.9	AD	DA1A 108120	9p13.3	***TPM2*** *190990*	Tropomyosin-2 (beta)	Sung et al (2003)	allelic to NEM4 and Cap disease (group 3), DA2B (group 16)
Arthrogryposis, distal, type 1B	16.10	AD	DA1B 614335	12q23.2	***MYBPC1*** *160794*	Myosin-binding proteinC, slow type	Gurnett et al (2010)	
Arthrogryposis, distal, type 2A, Freeman-Sheldon syndrome	16.11	AD	DA2A 193700	17p13.1	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al (2006)	
Arthrogryposis, distal, type 2B, Sheldon-Hall syndrome	16.12	AD	DA2B 601680	11p15.5	***TNNI2*** *191043*	Troponin I, fast skeletal	Sung et al (2003a) Kimber et al (2006)	
Arthrogryposis, distal, type 2B, Sheldon-Hall syndrome	16.13	AD	DA2B 601680	11p15.5	***TNNT3*** *600692*	Troponin T3, fast skeletal	Sung et al (2003)	
Arthrogryposis, distal, type 2B, Sheldon-Hall syndrome	16.14	AD	DA2B 601680	17p13.1	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al (2006)	
Arthrogryposis, distal, type 2B, Sheldon-Hall syndrome	16.15	AD	DA2B 601680	9p13.3	***TPM2*** *190990*	Tropomyosin-2 (beta)	Tajsharghi et al (2007) Ochala et al (2007)	allelic NEM4 and Cap disease (group 3), DA1A (group 16)
Arthrogryposis, distal, type 3	16.16	AD	DA3 114300	18p11.22-p11.21	***PIEZO2*** *613629*	Piezo-type mechanosensitive ion channe component 2	McMillin et al (2014)	
Arthrogryposis, distal, type 5	16.17	AD	DA5 108145	18p11.22-p11.21	***PIEZO2*** *613629*	Piezo-type mechanosensitive ion channe component 2	Coste et al (2013)	
Atrthrogryposis, Distal, type 7 (Trismus- pseudocamptodactyly syndrome)	16.18	AD	DA7 158300	17p13.1	***MYH8*** *160741*	Myosin heavy chain, 8, skeletal muscle, perinatal	Veugelers et al (2004) Toydemir et al (2006b)	

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Arthrogryposis multiplex congenita with nesprin-1 defect	16.19	AR	AMC	6q25.2	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (Nesprin-1)	Attali et al (2009)	allelic to EDMD4 (group 1) and SCAR8 (group 13)
Arthrogryposis multiplex congenita with cerebral and cerebellar atrophy	16.20	AR	LCCS7 616286	17q21.2	***CNTNAP1*** *602346*	Contactin-associated protein 1	Laquerriere et al (2014)	
Fetal akinesia deformation sequance with MUSK defect	16.21	AR	FADS 208150	9q31.3	***MUSK*** *601296*	Muscle-specific receptor tyrosine kinase	Tan-Sindhunata et al (2015)	allelic to CMS9 (group 11)
Fetal akinesia deformation sequance with DOK7 defect	16.22	AR	FADS 208150	4p16.3	***DOK7*** *610285*	Downstream of tyrosin kinase 7	Vogt et al (2009)	allelic to CMS10 (group 11)
Fetal akinesia deformation sequance with Rapsin defect	16.23	AR	FADS 208150	11p11.2	***RAPSN*** *601592*	Receptor-associated protein of the synapse, 43kD (Rapsyn)	Vogt et al (2008)	allelic to CMS11 (group 11)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1	16.24	AD	PEOA1 157640	15q26.1	***POLG*** *174763*	Polymerase, DNA, gamma (**M**)	Van Goethem et al (2001)	allelic to PEOB1 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2	16.25	AD	PEOA2 609283	4q35	***SLC25A4*** *103220*	Solute carrier family 25 (mitochondrial carrier, adenine nucleotide translocator), member 4 (**M**)	Kaukonen et al (2000)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3	16.26	AD	PEOA3 609286	10q24.31	***TWNK*** *606075*	Twinkle, mtDNA helicase (**M**)	Suomalinen et al (1997) Spelbrink et al (2001)	allelic to IOSCA (group 13)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4	16.27	AD	PEOA4 610131	17q23.3	***POLG2*** *604983*	Polymerase DNA, gamma 2 (**M**)	Longley et al (2006)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5	16.28	AD	PEOA5 613077	8q22.3	***RRM2B*** *604712*	Ribonucleotide reductase, M2 B (**M**)	Tyynismaa et al (2009)	allelic to MTDP8A (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 1	16.29	AR	PEOB1 258450	15q26.1	***POLG*** *174763*	Polymerase, DNA, gamma (**M**)	Deschauer et al (2007)	allelic to PEOA1 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	16.30	AR	PEOB2 616479	2p25.3	***RNASEH1*** *604123*	Ribonuclease H1 (**M**)	Reyes et al (2015)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3	16.31	AR	PEOB3 617069	16q21	***TK2*** *188250*	Thymidine kinase, mitochondrial (**M**)	Tyynismaa et al (2012)	allelic to MTDPS2 (group 16)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4	16.32	AR	PEOB4 617070	2p13.1	***DGUOK*** *601465*	Deoxyguanosine kinase (**M**)	Ronchi et al (2012)	
Mitochondrial DNA depletion myopathy	16.33	AR	MTDPS2 609560	16q21	***TK2*** *188250*	Thymidine kinase, mitochondrial (**M**)	Saada et al (2001)	allelic to PEOB3 (group 16)
Mitochondrial DNA depletion myopathy, encephalomyopathic form	16.34	AR	MTDPS5 612073	13q14.2	***SUCLA2*** *603921*	Succinate-CoA ligase, ADP-forming, beta subunit (**M**)	Elpeleg et al (2005)	
Mitochondrial DNA depletion myopathy	16.35	AR	MTDP8B 612075	8q22.3	***RRM2B*** *604712*	Ribonucleotide reductase, M2B (**M**)	Bourdon et al (2007)	allelic to PEOA5 (group 16)
Mitochondrial myopathy	16.36	AR		19p13.11	***SLC25A42*** *610823*	Solute carrier family 25, member 42 (**M**)	Shamseldin et al (2015)	
Progressive external ophthalmoplegia with optic atrophy, optic atrophy 1 with deafness	16.37	AD	125250	3q29	***OPA1*** *605290*	OPA1 protein (**M**)	Amati-Bonneau et al (2008) Hudson et al (2008)	
Mitochondrial myopathy and sideroblastic anemia 1	16.38	AR	MLASA1 600462	12q24.33	***PUS1*** *608109*	Pseudourine synthase 1 (**M**)	Bykhovskaya et al (2004) Fernandez-Vizarra (2007)	
Myopathy, lactic acidosis, and sideroblastic anemia-2	16.39	AR	MLASA2 613561	12p11.21	***YARS2*** *610957*	Tyrosyl-tRNA synthetase 2 (**M**)	Riley 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)
Mitochondrial myopathy	16.40	AD	IMMD 616209	22q11.23	***CHCHD10*** *615903*	Coiled-coil-helix-coiled-coil-helix domain containing 10 (**M**)	Ajroud-Driss et al (2014)	allelic to FTDALS2 and SMAJ (group 12)
Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis	16.41	AD	POIKTMP 615704	11q12.1	***FAM111B*** *615584*	Family with sequence similarity 111, member B	Mercier et al (2015)	
Combined oxidative phosphorylation deficiency 6;	16.42	XL	COXPD6 300816	Xq26.1	***AIFM1*** *300169*	Apoptosis-inducing factor, mitochondria-associated, 1 (**M**)	Morton et al (2017)	
Mitochondrial myopathy with severe neurological manifestations	16.43	AR		8q24.13	***TMEM65*** *616609*	Transmembrane protein 65	Nasli et al (2017)	
Mitochondrial complex IV deficiency	16.44	AR	220110	2q33.3	***FASTKD2*** *612322*	Fast kinase domains 2 (**M**)	Yoo et al (2017)	
Infantile-onset multisystem disease with progressive muscle weakness	16.45	AR	IMNEPD 616263	17q23.1	***PTRH2*** *608625*	Peptidyl-tRNA hydrolase 2	Hu et al (2014)	

NEW REFERENCES

GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES

Larson AA, Baker PR 2nd, Milev MP, Press CA, Sokol RJ, Cox MO, et al. TRAPPC11 and GOSR2 mutations associate with hypoglycosylation of α-dystroglycan and muscular dystrophy. Skelet Muscle. 2018 May 31;8(1):17. PMID: 29855340 [Items #2.40 and #2.41]

GROUP 3. CONGENITAL MYOPATHIES

Nilipour Y, Nafissi S, Tjust AE, Ravenscroft G, Hosseinejad Nedai H, Taylor R, et al. Ryanodine receptor type 3 (*RYR3*) as a novel gene associated with a myopathy with nemaline bodies. Eur J Neurol. 2018 Jun; 25(6):841–847. PMID: 29498452 [Item #3.13]

Majczenko K, Davidson AE, Camelo-Piragua S, Agrawal PB, Manfready RA, Li X, et al. Dominant mutation of CCDC78 in a unique congenital myopathy with prominent internal nuclei and atypical cores. Am. J. Hum. Genet. 2012; 91: 365–371. PMID:22818856 [Item #3.27]

Yuceyar, N., Ayhan, O., Karasoy, H., Tolun, A. Homozygous *MYH7* R1820W mutation results in recessive myosin storage myopathy: scapuloperoneal and respiratory weakness with dilated cardiomyopathy. Neuromuscul. Disord. 2015; 25: 340–344. PMID: 25666907 [Item #3.35]

Horstick, E.J., Linsley, J.W., Dowling, J.J., Hauser, M.A., McDonald, K.K., Ashley-Koch, A., et al. Stac3 is a component of the excitation-contraction coupling machinery and mutated in Native American myopathy. Nature Commun. 2013; 4:1952. PMID: 23736855 [Item #3.51]

GROUP 4. DISTAL MYOPATHIES

Bucelli RC, Arhzaouy K, Pestronk A, Pittman SK, Rojas L, Sue CM, et al. *SQSTM1* splice site mutation in distal myopathy with rimmed vacuoles. Neurology 2015; 85: 665–674. PMID:26208961 [Item #4.19]

Ruggieri A, Brancati F, Zanotti S, Maggi L, Pasanisi MB, Saredi S, et al. Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. Acta Neuropathol Commun. 2015 Jul 25; 3:44. PMID:26205529 [Item #4.20]

GROUP 5. OTHER MYOPATHIES

Hedberg C, Melberg A, Kuhl A, Jenne D, Oldfors A. Autosomal dominant myofibrillar myopathy with arrhythmogenic right ventricular cardiomyopathy 7 is caused by a *DES* mutation. Europ. J. Hum. Genet. 2012; 20: 984–985. PMID:22395865 [Item #5.12]

Kim HJ, Kim NC, Wang Y-D, Scarborough EA, Moore J, Diaz Z, et al. Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature 2013; 495: 467–473. PMID: 23455423 [Item #5.27]

GROUP 9. METABOLIC MYOPATHIES

Merezhinskaya N, Fishbein WN, Davis JI, Foellmer JW. Mutations in MCT1 cDNA in patients with symptomatic deficiency in lactate transport. Muscle Nerve 2000; 23: 90–97. PMID: 10590411 [Item #9.17]

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

Valdes-Mas R, Gutierrez-Fernandez A, Gomez J, Coto E, Astudillo A, Puente DA, et al. Mutations in filamin C cause

a new form of familial hypertrophic cardiomyopathy. Nature Commun. 2014; 5: 5326. PMID: 25351925 [Item #10.27]

Almomani R, Verhagen JMA, Herkert JC, Brosens E, van Spaendonck-Zwarts KY, Asimaki A, et al. Biallelic truncating mutations in *ALPK3* cause severe pediatric cardiomyopathy. J. Am. Coll. Cardiol. 2016; 67: 515–525. PMID: 26846950 [Item #10.28]

Papadopoulou LC, Sue CM, Davidson MM, Tanji K, Nishino I, Sadlock JE, et al. Fatal infantile cardioencephalomyopathy with COX deficiency and mutations in *SCO2*, a COX assembly gene. Nature Genet. 1999, 23: 333–337. PMID:10545952 [Item #10.35]

Antonicka H, Mattman A, Carlson CG, Glerum DM, Hoffbuhr KC, Leary SC, et al. Mutations in *COX15* produce a defect in the mitochondrial heme biosynthetic pathway, causing early-onset fatal hypertrophic cardiomyopathy. Am. J. Hum. Genet. 2003; 72: 101–114. PMID:12474143 [Item #10.36]

Li D, Parks SB, Kushner JD, Nauman D, Burgess D, Ludwigsen S, et al. Mutations of presenilin genes in dilated cardiomyopathy and heart failure. Am. J. Hum. Genet. 2006; 79: 1030–1039. PMID:17186461 [Item #10.56]

Carballo S, Robinson P, Otway R, Fatkin D, Jongbloed JDH, de Jonge N, et al. Identification and functional characterization of cardiac troponin I as a novel disease gene in autosomal dominant dilated cardiomyopathy. Circ. Res. 2009; 105: 375–382. PMID 19590045 [Item #10.67]

Peddy, S.B., Vricella, L.A., Crosson, J.E., Oswald, G.L., Cohn, R.D., Cameron, D.E., et al. Infantile restrictive cardiomyopathy resulting from a mutation in the cardiac troponin T gene. Pediatrics 117: 1830–1833, 2006. PMID: 16651346 [Item #10.87]

Purevjav E, Arimura T, Augustin S, Huby A-C, Takagi K, Nunoda S, et al. Molecular basis for clinical heterogeneity in inherited cardiomyopathies due to myopalladin mutations. Hum. Molec. Genet. 2012; 21: 2039–2053. PMID: 22286171 [Item #10.88]

Brodehl A, Ferrier RA, Hamilton SJ, Greenway SC, Brundler M-A, Yu W, et al. Mutations in *FLNC* are associated with familial restrictive cardiomyopathy. Hum. Mutat. 2016; 37: 269–279. PMID: 26666891 [Item #10.89]

Sasse-Klaassen S, Probst S, Gerull B, Oechslin E, Nurnberg P, Heuser A, et al. Novel gene locus for autosomal dominant left ventricular noncompaction maps to chromosome 11p15. Circulation 2004; 109: 2720–2723. PMID:15173023 [Item #10.93]

Vatta M, Mohapatra B, Jimenez S, Sanchez X, Faulkner G, Perles R, et al. Mutations in Cypher/ZASP in patients with dilated cardiomyopathy and left ventricular non-compaction. J. Am. Coll. Cardiol. 2003; 42: 2014–2017. PMID: 14662268 [Item #10.94]

Monserrat L, Hermida-Prieto M, Fernandez X, Rodriguez I, Dumont C, Cazon L, et al. Mutation in the alpha-cardiac actin gene associated with apical hypertrophic cardiomyopathy, left ventricular non-compaction, and septal defects. Europ. Heart J. 2007; 28: 1953–1961. PMID: 17611253 [Item #10.95]

Klaassen S, Probst S, Oechslin E, Gerull B, Krings G, Schuler P, et al. Mutations in sarcomere protein genes in left ventricular noncompaction. Circulation 2008; 117: 2893–2901. PMID: 18506004 [Item #10.96]

Luedde M, Ehlermann P, Weichenhan D, Will R, Zeller R, Rupp S, et al. Severe familial left ventricular non-compaction cardiomyopathy due to a novel troponin T (*TNNT2*) mutation. Cardiovasc. Res. 2010; 86: 452–460. PMID: 20083571 [Item #10.97]

Luxan G, Casanova JC, Martinez-Poveda B, Prados B, D'Amato G, MacGrogan D, et al. Mutations in the NOTCH pathway regulator MIB1 cause left ventricular noncompaction cardiomyopathy. Nature Med. 2013; 19: 193–201. PMID: 23314057 [Item #10.98]

Arndt A-K, Schafer S, Drenckhahn J-D, Sabeh MK, Plovie ER, Caliebe A, et al. Fine mapping of the 1p36 deletion syndrome identifies mutation of PRDM16 as a cause of cardiomyopathy. Am. J. Hum. Genet. 2013; 93: 67–77. PMID: 23768516 [Item #10.99]

Probst S, Oechslin E, Schuler P, Greutmann M, Boye P, Knirsch W, et al. Sarcomere gene mutations in isolated left ventricular noncompaction cardiomyopathy do not predict clinical phenotype. Circ. Cardiovasc. Genet. 2011; 4: 367–374. PMID: 21551322 [Items #10.100 and #10.101]

GROUP 11. CONGENITAL MYASTHENIC SYNDROMES

Engel AG, Selcen D, Shen XM, Milone M, Harper CM. Loss of MUNC13-1 function causes microcephaly, cortical hyperexcitability, and fatal myasthenia. Neurol Genet. 2016; 2:e105 PMID: 27648472 [Item #1.36]

Chaouch A, Porcelli V, Cox DM, Edvardson S, Scarcia P, de Grassi A, et al. Mutations in the mitochondrial citrate carrier SLC25A1 are associated with impaired neuromuscular transmission. J Neuromuscul Disord. 2014; 1:75–90 PMID:27648472 [Item #11.37]

Maselli RA, Vázquez J, Schrumpf L, Arredondo J, Lara M, Strober JB, et al. Presynaptic congenital myasthenic syndrome with altered synaptic vesicle homeostasis linked to compound heterozygous sequence variants in *RPH3A*. Mol Genet Genomic Med. 2018 Feb 14. PMID: 29441694 [Item #11.38]

GROUP 12. SPINAL MUSCULAR ATROPHIES MOTONEURON DISEASES

Smith BN, Topp SD, Fallini C, Shibata H, Chen H-J, Troakes C, et al. Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Sci. Transl. Med. 2017; 9: eaad9157. PMID: 28469040 [Item #12.60]

Brenner D, Muller K, Wieland T, Weydt P, Bohm S, Lule D, et al. *NEK1* mutations in familial amyotrophic lateral sclerosis. Brain 2016; 139: e28. PMID: 26945885 [Item #12.61]

Kenna KP, van Doormaal PT, Dekker AM, Ticozzi N, Kenna BJ, Diekstra FP, et al. *NEK1* variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genet. 2016; 48: 1037–1042. PMID: 27455347 [Item #12.61]

Iacomino M, Fiorillo C, Torella A, Severino M, Broda P, Romano C, et al. Spinal motor neuron involvement in a patient with homozygous PRUNE mutation. Eur J Paediatr Neurol. 2017 Dec 18. pii: S1090-3798(17)31863–9. PMID: 29307700 [Item #12.77]

GROUP 14. HEREDITARY MOTOR SENSORY NEUROPATHIES (HMSN)

Berciano J, Garcia A, Peeters K, Gallardo E, De Vriendt E, Palayo-Negro AL, et al. *NEFL* E396K mutation is associated with a novel dominant intermediate Charcot-Marie-Tooth disease phenotype. J. Neurol. 20154; 262: 1289–1300. PMID: 25877835 [Item #14.18]

Peeters K, Palaima P, Pelayo-Negro AL, Garcia A, Gallardo E, Garcia-Barredo et al. Charcot-Marie-Tooth disease type 2 G redefined by a novel mutation in *LRSAM1*. Ann. Neurol. 2016; 80: 823–833. PMID: 27686364 [Item #14.56]

Lassuthova P, Rebelo AP, Ravenscroft G, Lamont PJ, Davis MR, Manganelli F, et al. Mutations in *ATP1A1* cause dominant Charcot-Marie-Tooth type 2. Am. J. Hum. Genet. 2018; 102: 505–514. PMID: 29499166 [Item #14.68]

Polke JM, Laura M, Pareyson D, Taroni F, Milani M, Bergamin G, et al. Recessive axonal Charcot-Marie-Tooth disease due to compound heterozygous mitofusin 2 mutations. Neurology 2011; 77: 168–173. PMID: 21715711 [Item #14.69]

Nelis E, Erdem S, Van den Bergh PYK, Belpaire-Dethiou M-C, Ceuterick C, Van Gerwen V, et al. Mutations in *GDAP1*: autosomal recessive CMT with demyelination and axonopathy. Neurology 2002; 59: 1865–1872. PMID: 12499475 [Item #14.74]

Rebelo AP, Saade D, Pereira CV, Farooq A, Huff TC, Abreu L., et al. *SCO2* mutations cause early-onset axonal Charcot-Marie-Tooth disease associated with cellular copper deficiency. Brain. 2018 Mar 1;141(3):662–672. PMID: 29351582 [Item #14.84]

Rajadhyaksha, A.M., Elemento, O., Puffenberger, E.G., Schierberl, K.C., Xiang, J.Z., Putorti, M.L., et al. Mutations in FLVCR1 cause posterior column ataxia and retinitis pigmentosa. Am. J. Hum. Genet. 87: 643–654, 2010. PMID: 21070897 [Item #14.101]

Nahorski MS, Al-Gazali L, Hertecant J, Owen DJ, Borner GHH, Chen Y-C, et al. A novel disorder reveals clathrin heavy chain-22 is essential for human pain and touch development. Brain 2015; 138: 2147–2160. PMID: 26068709 [Item #14.102]

Habib AM, Matsuyama A, Okorokov AL, Santana-Varela S, Bras JT, Aloisi AM, et al. A novel human pain insensitivity disorder caused by a point mutation in *ZFHX2*. Brain 2018; 141: 356–376. PMID: 29253101 [Item #14.103]

GROUP 16. OTHER NEUROMUSCULAR DISORDERS

Shinwari JMA, Khan A, Awad S, Shinwari Z, Alaiya A, Alanazi M, et al. Recessive mutations in *COL25A1* are a cause of congenital cranial dysinnervation disorder. Am. J. Hum. Genet. 2015; 96: 147–152. PMID: 25500261 [Item #16.8]

Gurnett CA, Desruisseau DM, McCall K, Choi R, Meyer ZI, Talerico M, et al. Myosin binding protein C1: a novel gene for autosomal dominant distal arthrogryposis type 1. Hum. Molec. Genet. 2010; 19: 1165–1173. PMID: 20045868 [Item #16.10]

McMillin MJ, Beck AE, Chong JX, Shively KM, Buckingham KJ, Gildersleeve HIS, et al. Mutations in *PIEZO2* cause Gordon syndrome, Marden-Walker syndrome, and distal arthrogryposis type 5. Am. J. Hum. Genet. 2014; 94: 734–744. PMID: 24726473 [Item #16.16]

Coste B, Houge G, Murray MF, Stitziel N, Bandell M, Giovanni MA, et al. Gain-of-function mutations in the mechanically activated ion channel *PIEZO2* cause a subtype of distal arthrogryposis. Proc. Nat. Acad. Sci. 2013; 110: 4667–4672. PMID: 23487782 [Item #16.17]

Tan-Sindhunata MB, Mathijssen IB, Smit M, Baas F, de Vries JI, van der Voorn JP, et al. Identification of a Dutch founder mutation in *MUSK* causing fetal akinesia deformation sequence. Europ. J. Hum. Genet. 2015; 23: 1151–1157. PMID: 25537362 [Item #16.21]

Vogt J, Morgan NV, Marton T, Maxwell S, Harrison BJ. Beeson D, et al. Germline mutation in *DOK7* associated with fetal akinesia deformation sequence. J. Med. Genet. 2009; 46: 338–340. PMID: 19261599 [Item #16.22]

Vogt J, Harrison BJ, Spearman H, Cossins J, Vermeer S, ten Cate LN, et al. Mutation analysis of *CHRNA1, CHRNB1, CHRND*, and *RAPSN* genes in multiple pterygium syndrome/fetal akinesia patients. Am. J. Hum. Genet. 2008; 82: 222–227. PMID: 18179903 [Item #16.23]

Deschauer M, Tennant S, Rokicka A, He L, Kraya T, Turnbull DM, et al. MELAS associated with mutations in the *POLG1* gene. Neurology 2007; 68: 1741–1742. PMID: 17502560 [Item #16.29]

Reyes A, Melchionda L, Nasca A, Carrara F, Lamantea E, Zanolini A, et al. *RNASEH1* mutations impair mtDNA replication and cause adult-onset mitochondrial encephalomyopathy. Am. J. Hum. Genet. 2015; 97: 186–193. PMID: 26094573 [Item #16.30]

Tyynismaa H, Sun R, Ahola-Erkkila S, Almusa H, Poyhonen R, Korpela M, et al. Thymidine kinase 2 mutations in autosomal recessive progressive external ophthalmoplegia with multiple mitochondrial DNA deletions. Hum. Molec. Genet. 2012; 21: 66–75. PMID: 21937588 [Item #16.31]

Ronchi D, Garone C, Bordoni A, Gutierrez Rios P, Calvo SE, Ripolone M, et al. Next-generation sequencing reveals *DGUOK* mutations in adult patients with mitochondrial DNA multiple deletions. Brain 2012; 135: 3404–3415. PMID: 23043144 [Item #16.32]