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The 2014 version of the gene table of monogenic neuromuscular disorders (nuclear genome)

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General features

This table is published annually in the December issue. Its purpose is to provide the reader of *Neuromuscular Disorders* with an updated list of monogenic muscle 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 10-A (non-arrhythmogenic) and 10-B (arrhythmogenic); 11. Congenitalmyasthenic syndromes; 12. 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 clinical entities (phenotypic convergence due to genetic heterogenity, such as in CMT) and conversely a given clinical entity may be produced by a defect in several possible gene (phenotypic heterogeneity such as in LMNA defects) . 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 *Arrythmogenic 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.

Overview of the new data in the 2014 printed version

The updates comprise 27 new genes and 8 new phenotypic variants caused by a gene already listed in the

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

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

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

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2013 version. The list of added data is presented in Table 1. The 43 new key references are listed in pages 1109 to 1110 in this issue. Due to their increasing number **glycosylation defects** have been regrouped in a specific sub-category within group 2 (*congenital muscular dystrophies*)

Statistics extracted from the online full 2014 version (as of December 1 2013)

- 685 disease phenotypes
- 360 different genes (of which 28 code for a mitochondrial protein)
- 92 mapped loci awaiting gene identification
- 951 references

Citation of the gene table

- Printed version: Kaplan JC and Hamroun D The 2014 version of the gene table of neuromuscular disorders. Neuromuscul Disord. 23 (12), 1081–1111.
- Online version: GeneTable of Neuromuscular Disorders: *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 jean-claude.kaplan@inserm.fr.

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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.

Table 1

List of new material in the printed version

Updated group	Data and item # in the full list (see pages 1083 to 1109 in this issue)
1. Muscular dystrophies	New genes: *SMCHD1* (#1.10); *TNPO3* (#1.17); *TRAPPC11* (#1.37); *GMPPB* (#1.38) New phenotypes : LGMD2R/*DES* (#1.36); LGMD/*ISPD* (#1.41)
2. Congenital muscular dystrophies	New genes: *B3GNT1* (#2.24); *GMPPB* (#2.28 and # 2.35); *DPM1* (#2.31); *ALG13* (#2.33); *B3GALNT2* (#2.34); *TMEM5* (#2.36); *POMK* (#2.37) New phenotypes : L-CMD/LMNA (#2.15)
3. Congenital myopathies	New gene: *KLHL40* (#3.09); New phenotype: congenital myopathy/*PTPLA* (#3.38)
4. Distal myopathies	New phenotype: Welander syndrome/*TIA1* (#4.07)
5. Other myopathies	New phenotype: late onset axial myopathy/*RYR1* (#5.24)
9. Metabolic myopathies	New gene: *RBCK1* (#9.11)
11. Congenital myasthenic syndromes	New genes: *ALG2* (#11.24); *ALG14* (#11.25)
12. Motor neurone diseases	New genes: *SLC5A7* (#12.17); *UBA1* (#12.20); *BICD2* (#12.26); *EXOSC3* (#12.58); *HEXB* (#12.62)
14. Hereditary motor and sensory neuropathies	New genes: *GNB4* (#14.16); *HK1* (#14.26); *PDK3* (#14.34); *MARS* (#14.56); *SBF1* (#14.20); AIFM1 (#14.30); *TFG* (#14.55) New phenotypes: CMTR1C/*PLEKHG5* (#14.61); HMSN/*VRK1* (#14.75)

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Gene table of monogenic neuromuscular disorders (nuclear genome only)

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

Shaded background indicates newly added items.

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
GROUP 1. MUSCULAR DYSTROPHIES								
Duchenne muscular dystrophy; Becker muscular dystrophy	1.1	XR	DMD 310200 BMD 300376	Xp21.2	***DMD*** *300377*	Dystrophin	Monaco et al. (1986) Burghes et al. (1987) Koenig et al. (1987, 1988) Hoffman et al. (1987, 1988)	Allelic to CMD3B (group 10/A)
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	Xq27.2	***FHL1*** *300163*	Four and a half LIM domain 1	Gueneau et al. (2009)	Allelic to RSS (2), XPMA (5), XPMD (5) reducing body myopathy (group 5)
Emery-Dreifuss muscular dystrophy, autosomal dominant	1.4	AD	EDMD2 181350	1q21.2	***LMNA*** *150330*	Lamin A/C	Bonne et al. (1999) Worman and Bonne (2007)	Allelic to EDMD3 (group 1), LGMD1B (group 1), CMD1A (group 10/A), L-CMD (group 2), 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 181350	1q22	***LMNA*** *150330*	Lamin A/C	Raffaele di Barletta et al. (2000) Worman and Bonne (2007)	Allelic to EDMD2 (group 1), LGMD1B (group 1), L-CMD (group 2)CMD1A (group 10/A) 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	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Zhang et al. (2007)	Allelic to dilated cardiomyopathy with nesprin-1 defect, (group 10/A) SCAR8 (group 13), AMC with nesprin-1 defect (group 16)
Nesprin-2 related muscular dystrophy	1.7	AD	EDMD 5612999	14q23	***SYNE2*** *608442*	Spectrin repeat containing, nuclear envelope 2 (nesprin-2)	Zhang et al. (2007)	
LUMA related muscular dystrophy	1.8	AD		3p25.1	***TMEM43*** *612048*	Transmembrane protein 43 (=LUMA)	Liang et al. (2011)	Allelic to ARVD5 (group 10/A)

Table (*continued*)

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	1.9	AD	FSHMD1A 158900	4q35	***DUX4**** *606009* (**inappropriate reactivation*)	Double homeobox 4	Wijmenga et al. (1990, 1991, 1992, 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	1.10	AD	FSHMD1B 158901	18p11.32	***SMCHD1**** *(=KIAA0650) 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)	
Muscular dystrophy with generalized lipodystrophy	1.11	AD		17q21-q23	***PTRF*** *603198*	Polymerase I and transcript release factor (cavin-1) [*M*]	Hayashi et al. (2009)	
Limb girdle muscular dystrophies, dominant								
LGMD1A	1.12	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	1.13	AD	LGMD1B 159001	1q22	***LMNA*** *150330*	Lamin A/C	van der Koo et al. (1997) Muchir et al. (2000) Worman and Bonne (2007)	Allelic to EDMD2 (group 1), EDMD3 (group 1), CMD1A (group 10/A), CMT2B1 (group 14), [+ several other phenotypes FPLD2/ 151660, HGPS/176670, restrictive dermopathy/ 275210, not in this table]
LGMD1C	1.14	AD	LGMD1C 607801	3p25	***CAV3*** *601253*	Caveolin-3	Minetti et al. (1998) McNally et al. (1998)	Allelic to distal myopathy (group 4); hyper CKemia (group 5), RMD2 (group 6), CMH (group 10/A), LQT9 (group 10/A).
LGMD1D	1.15	AD	LGMD1D 603511	7q36.2	***DNAJB6*** *611332*	HSP-40 homologue subfamily B, number 6	Speer et al. (1999) Sarparanta et al. (2012) Harms et al. (2012)	
LGMD1E	1.16	AD	LGMD1E 602067	2q35	***DES*** *125660*	Desmin	Messina et al. (1997) Greenberg et al. (2012) Hedberg et al. (2012)	Allelic to myofibrillar myopathy (group 5), CMD1I (group 10/A), ARVD7 (group 10/B)
LGMD1F	1.17	AD	LGMD1F 608423	7q32.1-q32.2	***TNPO3*** *610032*	Transportin 3	Palenzuela et al. (2003), Melià et al. (2013), Torella et al. (2013)	
LGMD1G	1.18	AD	LGMD1G 609115	4q21	?		Starling et al. (2005)	
LGMD1H	1.19	AD	LGMD1H 613530	3p25.1-p23	?		Bisceglia et al. (2010)	
Limb girdle muscular dystrophies, recessive								
LGMD2A	1.20	AR	LGMD2A 253600	15q15.1	***CAPN3*** *114240*	Calpain -3	Beckmann et al. (1991) Young et al. (1992), Richard et al. (1995, 1997)	
LGMD2B	1.21	AR	LGMD2B 253601	2p13	***DYSF*** *603009*	Dysferlin	Bashir et al. (1994) Bashir et al. (1998) Liu et al. (1998)	Allelic to MM (group 4)

Table (*continued*)

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)
LGMD2C	1.22	AR	LGMD2C 253700	13q12	***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	1.23	AR	LGMD2D 608099	17q12-q21.33	***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)	
LGMD2E	1.24	AR	LGMD2E 604286	4q12	***SGCB*** *600900*	Beta-sarcoglycan	Lim et al. (1995) Bönnemann et al. (1995) Bönnemann et al. (1996)	
LGMD2F	1.25	AR	LGMD2F 601287	5q33	***SGCD*** *601411*	Delta-sarcoglycan	Passos-Bueno et al. (1996) Nigro et al. (1996)	Allelic to CMD1L (group 10/A)
LGMD2G	1.26	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/A)
LGMD2H	1.27	AR	LGMD2H 254110	9q31.2	***TRIM32*** *602290*	Tripartite motif-containing 32	Weiler et al. (1998) Frosk et al. (2002)	Allelic to sarcotubular myopathy (group 3)
LGMD2I	1.28	AR	LGMD2I (MDDGC5) 607155	19q13.3	***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	1.29	AR	LGMD2J 608807	2q31	***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/A), CMD1G (group 10/A)
LGMD2K	1.30	AR	LGMD2K (MDDGC1) 609308	9q34	***POMT1*** *607423*	Protein 0-mannosyltransferase 1	Balci et al. (2005) D'Amico et al. (2006)	Allelic to WWS (group 2)
LGMD2L	1.31	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	1.32	AR	LGMD2L (MDDGC4) 611588	9q31-q33	***FKTN*** *607440*	Fukutin	Murakami et al. (2006) Godfrey et al. (2006)	Allelic to FCMD (group 2) WWS (group 2) dilated cardiomyopathy (group 10/A)
LGMD2N	1.33	AR	LGMD2N (MDDGC2) 613158	14q24	***POMT2*** *607439*	Protein O-mannosyl transferase 2	Biancheri et al. (2007)	Allelic to WWS and MEB (group 2)
LGMD2O	1.34	AR	LGMD2O (MDDGC3) 613157	1p34	***POMGNT1*** *606822*	Protein O-linked mannose beta1,2-N-acetylglucosaminyl-transferase 1	Godfrey et al. (2007) Clement et al. (2008) Raducu et al. (2012)	Allelic to WWS and MEB (group 2)
LGMD2Q	1.35	AR	LGMD2Q 613723	8q24	***PLEC1*** *601282*	Plectin	Gundesli et al. (2010)	Allellic to MDEBS (group 5), and Myasthenic syndrome with plectin defect (group 11)
LGMD2R	1.36	AR	LGMD2R 615325	2q35	***DES*** *125660*	Desmin	Cetin et al. (2013)	Other desminopathies (items # 1.16, 5.2, 10.81, 10.35)
LGMD2S	1.37	AR	LGMD2S 615356	4q35.1	***TRAPPC11*** *614138*	trafficking protein particle complex 11	Bögershausen et al. (2013)	
LGMD2T = Limb-girdle muscular dystrophy with secondary dystroglycanopathy, related to GMPPB	1.38	AR	MDDGC14 615352	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	Carss et al. (2013)	MDDGA14, MDDGB14 (group 2)

(*Continued on next page*)

Table (*continued*)

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)
Recessive limb-girdle muscular dystrophy with primary alpha-dystroglycan defect	1.39	AR	MDDGC7 613818	3p21	***DAG1*** *128239*	Alpha-dystroglycan Dystroglycan 1 (dystrophin-associated glycoprotein 1)	Hara et al. (2011)	
Muscle dystrophy with glycosylation defect, type Io	1.40	AR	CDG1O 612937	1q22	***DPM3*** *605951*	Dolichyl-phosphate mannosyltransferase polypeptide 3	Lefeber et al. (2009)	
LGMD related to ISPD	1.41	AR		7p21.2	***ISPD*** *614631*	Isoprenoid synthase domain containing	Tasca et al. (2013)	Allelic to WWS/ MDDGA7 (2.12)
GROUP 2. CONGENITAL MUSCULAR DYSTROPHIES								
Congenital muscular dystrophy with merosin deficiency	2.1	AR	MDC1A 607855	6q2	***LAMA2*** *156225*	Laminin alpha2 chain of merosin (=laminin-2)	Tomé et al. (1994), Hillaire et al. (1994), Helbling-Leclerc et al. (1995), Allamand et al. (1997)	
Bethlem myopathy	2.2	AD	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	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	158810	2q37	***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 (recessive)	2.5	AR	158810	21q22.3	***COL6A2*** *120240*	Collagen type VI subunit alpha2	Gualandi et al. (2009)	Allelic to UCMD (group 2)
Ullrich syndrome	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 syndrome	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 syndrome	2.8	AR	UCMD 254090	2q37	***COL6A3*** *120250*	Collagen type VI subunit alpha 3	Demir et al. (2002)	Allelic toallelic to Bethlem myopathy (group 2) and myosclerosis (group 2)
Myosclerosis	2.9	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	2.10	AR	RSMD1 602771	1p36	***SEPN1*** *606210*	Selenoprotein N1	Moghadaszadeh et al. (1998, 2001), Ferreiro et al. (2002a, 2004)	Allelic to CFTD (group 3), multiminicore disease (group 3), and desminrelated myopathy with Mallory bodies (group 5)
Rigid spine syndrome	2.11	AR	RSMD1 602771	Xq26.3	***FHL1 300163***	Four and a half LIM domain 1	Shalaby et al. (2008)	Allelic to EDMD6 (1), RSS (2), XPMA(5), XPMD (5)
Congenital muscular dystrophy with integrin defect	2.12	AR		12q13	***ITGA7*** *600536*	Integrin α7	Hayashi et al. (1998)	
Congenital muscular dystrophy with dynamin 2 defect	2.13	AD		19p13.2	***DNM2 602378***	Dynamin 2	Susman et al. (2008)	Allelic to CNM (group 3)
Congenital musuclar dystrophy with telethonin defect	2.14	AR		17q12	***TCAP*** *604488*	Titin-cap (telethonin)	Ferreiro et al. (2011)	Allelic to LGMD2G (group 1)
Congenital muscular dystrophy due to LMNA defect (L-CMD)	2.15	AD		1q21.2	***LMNA*** *150330*	Lamin A/C	Quijano-Roy et al. (2005)	Allelic to EDMD3 (group 1), LGMD1B (group 1), L-CMD (group 2), CMD1A (group 10/A), CMT2B1 (group 14), [+ several other phenotypes not in this table: FPLD2/ 151660, HGPS/176670, restrictive dermopathy/ 275210, MADA/248370]
Congenital muscle dystrophies due to defective glycosylation								
Fukuyama congenital muscular dystrophy	2.16	AR	MDDGA4 MDDGB4 253800	9q31-q33	***FKTN*** *607440*	Fukutin	Toda et al. (1993), Kobayashi et al. (1998)	Allelic to WWS (group 2)

Table (*continued*)

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)
Walker-Warburg syndrome (WWS)	2.17	AR	MDDGA4 MDDGB4 253800	9q31-q33	***FKTN*** *607440*	Fukutin	Beltran-Valero de Bernabe (2003), Mercuri et al. (2009)	Allelic to LGMD2L (group 1) and Fukuyama (group 2)
Walker-Warburg syndrome (WWS)	2.18	AR	MDDGA1 MDDGA1 236670	9q34	***POMT1*** *607423*	Protein-O-mannosyltransferase 1	Beltran-Valero De Bernabe et al. (2002), van Reeuwijk et al. (2006), Mercuri et al. (2009)	Allelic to LGMD2K (group 1)
Walker-Warburg syndrome (WWS)	2.19	AR	MDDGA2 MDDGA2 613150	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)	2.20		MDDGA5 MDDGB5 613153	19q13	***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)	2.21	AR	MDDGA2 MDDGA2 253280	1p34.1	***POMGNT1*** *606822*	O-mannose beta1,2-*N*-acetylglucosaminyl transferase	Taniguchi et al. (2003), Mercuri et al. (2009)	Allelic to MEB (group 2)
Walker-Warburg syndrome (WWS)	2.22	AR	MDDGA7 614643	7p21.2	***ISPD*** *614631*	Isoprenoid synthase domain containing	Roscioli et al. (2012), Willer et al. (2012)	
Walker-Warburg syndrome (WWS)	2.23	AR	MDDGA8 614830	3p22.1	***GTDC2*** *614828*	Glycosyltransferase-like domain containing 2	Manzini et al. (2012)	
Walker-Warburg syndrome (WWS)	2.24	AR	MDDGA13 615287	11q13.2	***B3GNT1*** *605517*	UDP-GlcNAc:betaGal beta-1,3-*N*-acetylglucosaminyl-transferase 1	Buysse et al. (2013), Shaheen et al. (2013	
Muscle–eye–brain disease (MEB)	2.25	AR	MDDGA2 253280	1p34.1	***POMGNT1*** *606822*	O-mannose beta1,2-*N*-acetylglucosaminyl transferase	Yoshida et al. (2001), Taniguchi et al. (2003)	Allelic to WWS (group 2)
Muscle–eye–brain disease (MEB)	2.26	AR	MEB MDDGA5 MDDGB5 613153	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)	2.27	AR	MEB MDDGA2 MDDGA2 613150	14q24.3	***POMT2*** *607439*	Protein O-mannosyl transferase 2		Allelic to WWS (group 2)
Muscle–eye–brain disease	2.28	AR	MDDGC14 615350	3p21.31	***GMPPB*** *615320*	GDP-mannose pyrophosphorylase B	de Carss et al. (2013)	Allelict to LGMD2T (group 1) and, to MDDGB14 (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.29	AR	MDDGA5 MDDGB5 613153	19q13	***FKRP*** *606596*	Fukutin related protein	Brockington et al. (2001b), Topaloglu et al. (2003), Mercuri et al. (2009)	Allelic to LGMD2I (group 1), WWS (group 2), MEB (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.30	AR	MDDGA6 MDDGB6 613154	22q12	***LARGE*** *603590*	Like-glycosyl transferase	Longman et al. (2003), Mercuri et al. (2009)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.31	AR	CDG1E 608799	20q13.13	***DPM1*** *603503*	Dolichyl-phosphate mannosyltransferase 1, catalytic subunit	Yang et al. (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan and severe epilepsy	2.32	AR	CDG1U 615042	9q34.13	***DPM2*** *603564*	Dolichyl-phosphate mannosyltransferase polypeptide 2, regulatory subunit	Barone et al. (2012)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan	2.33	XR	CDG1S 300884	Xq23	***ALG13*** *300776*	UDP-*N*-acetylglucosaminyltransferase subunit	Timal et al. (2012	Congenital myasthenia Group 11…
Congenital muscular dystrophy with hypoglycosylation of dystroglycan WWWS/MEB like	2.34	AR	MDDGA11 615181	1q42.3	***B3GALNT2*** *610194*	Beta-1,3-*N*-acetylgalactosaminyltransferase 2	Stevens et al. (2013)	

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Table (*continued*)

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Congenital muscular dystrophy with hypoglycosylation of dystroglycan and mental retardation	2.35	AR	MDDGB14 615351	3p21.31	*GMPPB 615320*	GDP-mannose pyrophosphorylase B	Carss et al. (2013)	Allelit to LGMD2T (group 1), MDDGC14 (group 2)
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A10	2.36	AR	MDGGA10 615041	12q14.2	***TMEM5*** *605862*	Transmembrane protein 5	Vuillaumier-Barrot et al. (2013)	
Congenital muscular dystrophy with hypoglycosylation of dystroglycan type A,12	2.37	AR	MDDGA12 615249	8p11.21	***POMK (=SGK196)***	Protein-*O*-mannose kinase	Jae et al. (2013)	
Other congenital muscular dystrophies								
Congenital muscle dystrophy with joint hyperlaxity	**2.38**	AR		3p23–21	*?*		Tetreault et al. (2006)	
Congenital muscle dystrophy with mitochondrial structural abnormalities (megaconial type)	**2.39**	AR	MDCMC 602541	22q13	***CHKB*** *612395*	Choline kinase beta	Mitsuhashi et al. (2011)	
Congenital muscular dystrophy,	2.40	AR	MDC1B 604801	1q42	*?*		Brockington et al. (2000)	
GROUP 3. CONGENITAL MYOPATHIES								
Nemaline myopathy	3.1	AD	NEM1 609284	1q21.2	***TPM3*** *191030*	Tropomyosin 3	Laing et al. (1992) Laing et al. (1995b) Tan et al. (1999) Wattanasirichaigoon et al. (2002) [add here, already in the list]	
Nemaline myopathy	3.2	AR	NEM2 256030	2q22	***NEB*** *161650*	Nebulin	Wallgren-Pettersson et al. (1995, 2002) Pelin et al. (1999) Lehtokari et al. (2006)	
Nemaline myopathy	3.3	AD	NEM3 161800	1q42.1	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Nowak et al. (1999)	Allelic to CFTD (group 3)
Nemaline myopathy	3.4	AD	NEM4 609285	9p13	***TPM2*** *190990*	Tropomyosin 2 (beta)	Donner et al. (2002)	See Item #3.5
Nemaline myopathy with Escobar syndrome	3.5	AR	26500	9p13	***TPM2*** *190990*	Tropomyosin 2 (beta)	Monnier et al. (2009)	See Item #3.4
Nemaline myopathy	3.6	AR	NEM5 605355	19q13	***TNNT1*** *191041*	Troponin T type 1 (skeletal, slow)	Johnston et al. (2000)	
Nemaline myopathy	3.7	AD	NEM6 609273	15q22.31	***KBTBD13*** *613727*	Kelch repeat and BTB (POZ) domain containing 13	Gommans et al. (2003) Samburghin et al. (2010)	
Nemaline myopathy	3.8	AR	NEM7 610687	14q12	***CFL2*** *601443*	*Cofilin 2 (muscle)*	Agrawal et al. (2007)	
Severe autosomal-recessive nemaline myopathy	3.9	AR	NEM8 615348	2p22.1	***KLHL40*** *615340*	Kelch-like family member 40	Ravenscroft et al. (2013)	
Myopathy, congenital, with fiber-type disproportion	3.10	AD	CFTD 255310	1q42.1	***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	3.11	AR	CFTD 255310	1p36	***SEPN1*** *606210*	Selenoprotein N1	Clarke et al. (2006)	Allelic to RSMD1 (group 2), multiminicore disease (group 3), desmin-related myopathy with Mallory bodies (group 5)

Table (*continued*)

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Myopathy, congenital, with fiber-type disproportion	3.12	AD	CFTD 255310	1q21.2	**TPM3** *191030*	Tropomysosin 3	Clarke et al. (2008)	Allelic to NEM1 (group 3)
Myopathy, congenital, with fiber-type disproportion	3.13	AR	CFTD 255310	19q13.1	***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	3.14	AD	CFTD 255310	14q12	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, b	Ortolano et al. (2011)	Allelic to MPD1 (group 4), CMH1 (group 10/A), CMD1S (group 10/A)
Myotubular myopathy	3.15	XR	MTM1 310400	Xq28	***MTM1*** *300415*	Myotubularin 1	Thomas et al. (1987), Laporte (1996, 1997, 2000)	
Centronuclear myopathy, dominant	3.16	AD	CNM 160150	19.p13.2	***DNM2*** *602378*	Dynamin 2	Bitoun et al. (2005)	
Centronuclear myopathy, recessive	3.17	AR	255200	2q14	***BIN1*** *601248*	Amphiphysin	Nicot et al. (2007)	
Centronuclear myopathy, recessive	3.18	AR	255200	19q13.1	***RYR1*** *180901*	Ryanodine receptor	Wilmshurst et al. (2010)	Allelic to CCD (group 3), minicore myopathy with external ophtalmoplegia (group 3), MHS1 (group 8)
Central core disease, dominant	3.19	AD	CCD 117000	19q13.1	***RYR1*** *180901*	Ryanodine receptor	Kausch et al. (1991) Zhang et al. (1993) Quane et al. (1993), Robinson et al. (2002)	Allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Central core disease, recessive (transient multiminicore myopathy)	3.20	AR	CCD 117000	19q13.1	***RYR1*** *180901*	Ryanodine receptor	Ferreiro et al. (2002a), Jungbluth et al. (2002)	Allelic to CCD (group 3), minicore myopathy with external ophthalmoplegia (group 3), MHS1 (group 8)
Multiminicore disease with external ophtalmoplegia	3.21	AR	255320	19q13.1	***RYR1*** *180901*	Ryanodine receptor	Monnier et al. (2003), Jungbluth et al. (2005)	Allelic to CCD (group 3), CCD (group 3), MHS1 (group 8)
Multiminicore disease, classical form	3.22	AR	255320	1p36	***SEPN1*** *606210*	Selenoprotein N1	Ferreiro et al. (2002b), 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	3.23	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	multiple EGF-like domains 10	Logan et al. (2011)	See Item #3.23
Recessive congenital myopathy with minicores	3.24	AR	EMARDD 614399	5q23.2	***MEGF10*** *612453*	multiple EGF-like domains 10	Boyden et al. (2012)	See Item #3.22
Hyaline body myopathy (recessive)	3.25	AR	255160	3p22.2-p21.32	?	?	Onengut et al. (2004)	
Hyaline body myopathy, dominant (myosin storage myopathy)	3.26	AD	608358	14q12	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, b	Tajsharghi et al. (2003), Bohlega et al. (2004) Laing et al. (2005)	Allelic to CFTD (group 3), MPD1 (group 4), CMH1 (group 10/A), CMD1S (group 10/A)
Myosin storage myopathy and cardiomyopathy, recessive	3.27	AR		14q12	***MYH7*** *160780*	Myosin, heavy chain 7, cardiac muscle, b	Tajsharghi et al. (2007a)	Allelic to CFTD (group 3), MPD1 (group 4), CMH1 (group 10/A), CMD1S (group 10/A)
Myosin lia myopathy dominant (inclusion body myopathy)	3.28	AD	IBM3 605637	17p13.1	***MYH2*** *160740*	Myosin, heavy chain 2, skeletal muscle, adult	Martinsson et al. (1999, 2000)	
Myosin lia myopathy, recessive	3.29	AR		17p13.1	***MYH2*** *160740*	Myosin, heavy chain 2, skeletal muscle, adult	Tajsharghi et al. (2010)	
Cap myopathy	3.30	AD	CAPM2 190900	9p13	***TPM2*** *190990*	Tropomyosin 2, b	Tajsharghi et al. (2007) Lehtokari et al. (2007)	Allelic to NEM4 (group 3) DA1 (group 16) and DA2B (group 16)

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Cap myopathy	3.31	AD	CAPM1 609284	1q21.2	**TPM3** *191030*	Tropomysosin 3	De Paula et al. (2009) Ohlsson et al. (2009)	Allelic to NEM1 (group 3)
Cap myopathy	3.32	AD		1q42.1	***ACTA1*** *102610*	Actin, alpha 1, skeletal muscle	Hung et al. (2010)	Allelic to NEM3 (group 3)
Congenital neuromuscular disease with uniform type 1 fiber	3.33	AD	CUMDU1 117000	19q13.1	***RYR1*** *180901*	Ryanodine receptor I	Sato et al. (2007)	Allelic to CDD (group 3), multi-minicore disease (group 3), MHS1 (group 8)
Congenital myopathy with fatal cardiomyopathy	3.34	AR	EOMFL 611705	2q31	***TTN*** *188840*	Titin	Carmignac et al. (2007)	Allelic to LGMD2 J (group 1), TMD (group 4) HMERF (group 5), CMH9 (group 10/A), CMD1G (group 10/A)
Congenital skeletal myopathy and fatal cardiomyopathy	3.35	AR		11p11.2	***MYBPC3*** *600958*	Cardiac myosin binding protein-C	Tajsharghi et al. (2010)	Allelic to CMH4 (group 10/A)
Compton-North congenital myopathy	3.36	AR		12q11-q12	***CNTN1*** *600016*	Contactin-1	Compton et al. (2008)	
Sarcotubular myopathy	3.37	AR		9q31	***TRIM32*** *602290*	Tripartite motif containing 32 (ubiquitin ligase)	Schoser et al. (2005)	Allelic to LGMD2H (group 1)
Congenital myopathy related to PTPLA	3.38	AR		10p12.33	***PTPLA (=HACD1)*** *610467*	Protein tyrosine phosphatase-like (3-Hydroxyacyl-CoA dehydratase)	Muhammad et al. (2013)	ARVD6 (item #10–80)
GROUP 4. DISTAL MYOPATHIES								
Distal recessive myopathy (Miyoshi)	4.1	AR	MM 254130	2p12–14	***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	***TTN*** *188840*	Titin	Haravuori et al. (1998) Haravuori et al. (2001) Hackman et al. (2002)	Allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), HMERF (group 5), CMH9 (group 10/A), CMD1G (group 10/A)
Distal myopathy with rimmed vacuoles (Nonaka) and Hereditary inclusion body myopathy	4.3	AR	NM 605820 IBM2 600737	9p12-p12	***GNE*** *603824*	Glucosamine (UDP-N-acetyl)-2-epimerase/ N-acetylmannosamine kinase	Mitrani-Rosenbaum et al. (1996), Ikeuchi et al. (1997), Eisenberg et al. (2001)	
Distal myopathy (Laing)	4.4	AD	MPD1 160500	14q11.2	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Laing et al. (1995a), Mastaglia et al. (2000), Meredith et al. (2004)	Allelic to CFTD (group 3), myosin storage myopathy (group 3), CMH1 (group 10/A), CMD1S (group 10/A)
Vocal cord and pharyngeal distal myopathy	4.5	AD	MPD2 (VCPDM) 606070	5q31	*?*		Feit et al. (1998)	
Adult onset distal myopathy	4.6	AD	MPD3 610099	8p22-q11	*?*		Haravuori et al. (2004)	
Welander distal myopathy	4.7	AD	WDM 604454	2p13	***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	19.p13	*?*		Servidei et al. (1999)	
Distal myopathy with myotilin defect	4.9	AD		5q31	***MYOT*** *(=TTID) 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		2q22	***NEB*** *161650*	Nebulin	Wallgren-Pettersson et al. (2007)	Allelic to NEM2 (group 3)
Distal myopathy with caveolin defect	4.11	AD		3p25	***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 (group 10/A)

Table (*continued*)

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Late onset distal myopathy (Markesbery-Griggs)	4.12	AD		10q22	***LDB3*** *605906*	ZASP	Griggs et al. (2007)	Allelic to MFM (group 5)
Early onset calf distal myopathy	4.13	AR		11p14–12	***AN05*** *(TMEM16E)* *608662*	Anoctamin 5	Bolduc et al. (2010)	Allelic to LGMD2L (group 1)
Early onset distal myopathy with KLHL9 defect	4.14	AD		9p22	***KLHL9*** *611201*	Kelch-like 9 (Drosophila)	Cirak et al. (2010)	
Dynamin 2 related distal myopathy	4.15	AD	CNM 160150	19.p13.2	***DNM2*** *602378*	Dynamin 2	Fischer et al. (2006)	Allelic to CNM (group 3) and CMTDIB (group 14)
Filamin C related distal myopathy	4.16	AD	MPD4 614065	7q32	***FLNC*** *102565*	Filamin C, gamma (actin binding protein 280)	Duff et al. (2011)	Alleic to MFM5 (group 5)
Distal myopathy with VCP defect	4.17	AD	IBMPFD 167320	9p13-p12	***VCP*** *601023*	Valosin-containing protein	Palmio et al. (2011)	Allelic to IBMBFD (group 5)
GROUP 5. OTHER MYOPATHIES								
A. Myofibrillar myopathies								
Myofibrillar myopathy, alpha-B crystallin related	5.1	AD	MFM 608810	11q22	***CRYAB*** *123590*	Crystallin, alpha B	Vicart et al. (1998), Selcen et al. (2003)	
Myofibrillar myopathy, desmin-related myopathy	5.2	AD	DRM 601419	2q35	***DES*** *125660*	Desmin	Goldfarb et al. (1998) Munoz-Marmol et al. (1998)	
Desmin-related myopathy with Mallory bodies	5.3	AD	602771	1p36	***SEPN1*** *606210*	Selenoprotein N1	Ferreiro et al. (2004)	Allelic to RSMD1 (group 2), CFTD (group 3) multiminicore disease (group 3)
Myofibrillar myopathy	5.4	AD	MFM 609452	10q22	***LDB3*** 605906	ZASP	Selcen and Engel (2005)	Allelic to Markesbery-Griggs (group 4) and to CMD1C (group 10/A)
Myofibrillar myopathy with arrhythmogenic right ventricular cardiomyopathy	5.5	AD	MFM/ARVC 609160	10q22	?		Melberg et al. (1999) Kuhl et al. (2008)	
Myofibrillar myopathy, myotilin related	5.6	AD	MFM 609200	5q31	***MYOT*** *(=TTID)* *604103*	Myotilin (titin immunoglobulin domain protein)	Selcen and Engel (2004)	Allelic to LGMD1A (group 1), spheroid body myopathy (group 5)
Spheroid body myopathy	5.7	AD	182920	5q31	***MYOT*** *(=TTID)* *604103*	Myotilin (titin immunoglobulin domain protein)	Foroud et al. (2005)	Allelic to LGMD1A (group 1), MFM (group 5)
Myofibrillar myopathy, filamin-C related	5.8	AD	MFM 5609524	7q32	***FLNC*** *102565*	Filamin C, gamma (actin binding protein 280)	Vorgerd et al. (2005)	Allelic to filamin C related distal myopathy (group 4)
Myofibrillar myopathy with BAG3 defect	5.9	AD		10q25-q26	***BAG3*** *603883*	BCL2-associated athanogene 3	Selcen et al. (2009)	
B. Miscellaneous								
Danon disease	5.10	XD	GSD IIb 300257	Xq24	***LAMP2*** *309060*	Lysosomal-associated membrane protein 2	Nishino et al. (2000) Musumeci et al. (2005)	
Myopathy with excessive autophagia	5.11	XR	MEAX (XMEA) 310440	Xq28			Saviranta et al. (1988) Villard et al. (2000) Minassian et al. (2002), Munteanu et al. (2008)	
Oculopharyngeal muscular dystrophy	5.12	AD	OPMD 164300	14q11.2-q13	***PABPN1*** *602279*	Poly(A) binding protein, nuclear 1	Brais et al. (1995, 1998) Robinson et al. (2005)	
Hereditary myopathy with early respiratory failure (Edström myopathy)	5.13	AD	HMERF 603689	2q24-3	***TTN*** *188840*		Nicolao et al. (1999) Lange et al. (2005)	Allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), CMH9 (group 10/A), CMD1G (group 10/A)

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Epidermolysis bullosa simplex associated with late-onset muscular dystrophy	5.14	AR	MDEBS 226670	8q24-qter	***PLEC1*** *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	5.15	AR		2q32	***GDF8*** *601788*	Growth differentiation factor 8 (myostatin)	Schuelke et al. (2004)	
Fibrodysplasia ossificans progressiva	5.16	AD	FOP 135100	2q23-q24	***ACVR1*** *102576*	Activin A receptor, type 1	Shore et al. (2006)	
HyperCKemia, idiopathic	5.17	AD	123320	3p25	***CAV3*** *601253*	Caveolin-3	Carbone et al. (2000)	Allelic to LGMD1C (group 1) and RMD2 (group 6), CMH (group 10/A)
X-linked myopathy with postural muscle atrophy	5.18	XR	XMPMA 300696	Xq26.3	***FHL1*** *300163*	Four and a half LIM domain 1	Windpassinger et al. (2008)	Allelic to Emery-Dreifuss MD X-linked type 2 (group 1) reducing body myopathy (group 3), XPMD (group 5)
Scapuloperoneal myopathy	5.19	XD	XPMD 300695	Xq26.3	***FHL1*** *300163*	Four and a half LIM domain 1	Quinzil et al. (2008)	Allelic to Emery-Dreifuss MD X-linked type 2 (group 1) reducing body myopathy (group 3), XMPMA (group 5)
Reducing body myopathy	5.20	XD	300717 300718	Xq26.3	***FHL1*** *300163*	Four and a half LIM domain 1	Schessl et al. (2008), Shalaby et al. (2009)	Allelic to EDMD6 (group 1), RSS (group 2), XPMA and XPMD (group 5)
Episodic muscle weakness, X-linked	5.21	XR	EMWX 300211	Xp22.3	*?*		Ryan et al. (1999)	
Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia	5.22	AD	IBMPFD 167320	9p13-p12	***VCP*** *601023*	Valosin-containing protein	Watts et al. (2004), Haubenberger et al. (2005)	Allelic to distal myopathy with VCP defect (group 4)
Myopathy with exercise intolerance, Swedish type	5.23	AR	HML 255125	12q24.1	***ICSU*** *611911*	Iron–sulfur cluster scaffold homolog (*E. coli*)	Mochel et al. (2008)	
Late onset axial myopathy related to RYR1	5.24	AD		19q13.1	***RYR1*** *180901*	Ryanodine receptor 1 (skeletal)	Løseth et al. (2013)	Allelic to items 3.12, 3.17, 3.18, 3.19, 3.20, 3.32
GROUP 6. MYOTONIC SYNDROMES								
Myotonic dystrophy (Steinert)	6.1	AD	DM11 60900	19q13	***DMPK*** *605377*	Dystrophia myotonica-protein kinase	Renwick et al. (1971) Friedrich et al. (1987) Harley et al. (1992) Buxton et al. (1992) Aslanidis et al. (1992) Mahadevan et al. (1992) Fu et al. (1992) Brook et al. (1992)	
Myotonic dystrophy type 2 (proximal myotonic myopathy)	6.2	AD	DM2 (PROMM) 602668	3q21	***ZNF9*** *116955*	Zinc finger protein 9	Ranum et al. (1998) Liquori et al. (2001)	
Myotonia, dominant (Thomsen)	6.3	AD	*See under Ion channel muscle diseases*					
Myotonia, recessive (Becker)	6.4	AR	*See under Ion channel muscle diseases*					
Rippling muscle disease, dominant	6.5	AD	RMD1 600332	1q41	*?*		Stephan et al. (1994)	
Rippling muscle disease, dominant	6.6	AD	RMD2 606072	3p25	***CAV3*** *601253*	Caveolin-3	Betz et al. (2001)	Allelic to LGMD1C (group 1), hyper- Ckemia (group 5), RMD2 (group 6), CMH (group 10/A)
Rippling muscle disease, recessive	6.7	AR	RMD2 606072	3p25	***CAV3*** *601253*	Caveolin-3	Kubisch et al. (2003, 2005)	Allelic to LGMD1C (group 1), hyper- Ckemia (group 5), RMD2 (group 6), CMH (group 10/A)

Table (*continued*)

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)
Schwartz-Jampel syndrome	6.8	AR	SJS1 255800	1p34-p36.1	***HSPG2*** *142461*	Heparan sulfate proteoglycan 2 (perlecan)	Nicole et al. (1995, 2000)	
Brody disease	6.9	AR AD	601003	16p12	***ATP2A1= SERCA1*** *108730*	ATPase, Ca++ transporting, cardiac muscle, fast twitch 1	Odermatt et al. (1996)	
GROUP 7. ION CHANNEL MUSCLE DISEASES								
(A) Chloride channel								
Myotonia congenita, dominant (Thomsen)	7.1	AD	THD 160800	7q35	***CLCN1*** *118425*	Muscle chloride channel	Koch et al. (1992b) George Jr et al. (1993)	Allelic to Becker myotonia (group 7)
Myotonia, recessive (Becker)	7.2	AR	255700	7q35	***CLCN1*** *118425*	Muscle chloride channel	Koch et al. (1992b)	Allelic to Thomsen myotonia (group 7)
(B) Sodium channel								
Hyperkalaemic periodic paralysis	7.3	AD	hyperKPP 170500	17q23	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha	Fontaine et al. (1990), Ptáček et al. (1991a), Rojas et al. (1991), Miller et al. (2004)	Allelic to HOKPP2 (group 7), PMC (group 7), K-aggravated myotonia (group 7)
Hypokalaemic periodic paralysis, type 2	7.4	AD	hypoKPP 170400	17q23	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha	Bulman et al. (1999) Jurkat-Rott et al. (2000)	Allelic to HYPP (group 7), PMC (group 7), K-aggravated myotonia (group 7)
Paramyotonia congenita	7.5	AD	PMC 168300	17q23	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha	Ptáček et al. (1991b, 1992a, 1993), Ebers et al. (1991) Koch et al. (1992a) Mc Clatchey et al. (1992)	Allelic to HYPP (group 7), HOKPP2 (group 7), K-aggravated myotonia (group 7)
Potassium-aggravated myotonia	7.6	AD	608390	17q23	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha	Ptáček et al. (1992a, 1992b, 1994a), Heine et al. (1993) Lerche et al. (1993)	Allelic to HYPP (group 7.3), HOKPP2 (group 7.4), PMC (group 7.5)
Long QT syndromes	7.7	*See under Hereditary cardiomyopathies (group 10-B, online only)*						
(C) Calcium channel								
Hypokalaemic periodic paralysis, type 1	7.8	AD	hypoKPP1 170400	1q31-q32	***CACNA1S*** *(ex* ***CACNL1A3****)* *114208*	Calcium channel, voltage-dependent, L type, alpha 1S subunit	Fontaine et al. (1994) Ptáček et al. (1994b) Jurkat-Rott et al. (1994) Elbaz et al. (1995)	
Acetazolamide responsive hereditary paroxysmal cerebellar ataxia (also listed in group 13 "Ataxias")	7.9	AD	APCA 108500	19p13	***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	7.10	AD	EA2 108500	19p13	***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)
(D) Potassium channel								
Hypokalaemic periodic paralysis, type 3	7.11	AD	hypoKPP3 170400	11q13	***KCNE3*** *604433*	Potassium voltage-gated channel, Isk-related family, member 3	Abbott et al. (2001)	
Episodic ataxia/ myokymia	7.12	AD	EA1 160120	12p13	***KCNA1*** *176260 (voltage gated K+ channel)*	Potassium voltage-gated channel, shaker-related subfamily, member 1	Browne et al. (1994), Adelman et al. (1995)	
Thyrotoxic hypokalemic periodic paralysis	7.13		TTPP2 613239	17p11.2	***KCNJ18*** *613239*	Kir2.6 (inwardly rectifying potassium channel 2.6)	Ryan et al. (2010)	
Periodic paralysis, potassium sensitive cardiodysrythmic (Andersen's syndrome)	7.14	*See LQ7 under hereditary cardiomyopathies (group 10-B online only)*						
Long QT syndromes	7.15	*See under hereditary cardiomyopathies (group 10-B online only)*						

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Table (*continued*)

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 8. MALIGNANT HYPERTHERMIAS								
Malignant hyperthermia	8.1	AD	MHS1 145600	19q13.1	***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	1q31-q32	***CACNA1S*** *(ex* ***CACNL1A3****) 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								
(A) Glycogen storage diseases								
Glycogen storage disease type II (Pompe disease) also listed in group 10 *Hereditary cardiomyopathies*	9.1	AR	GSDII 232300	17q25	***GAA*** *606800*	Glucosidase, alpha; acid	Hers (1963), Martiniuk et al. (1990), Wokke et al. (1995)	Allelic to GSDII (group 10/A)
Glycogen storage disease type IIIa	9.2	AR	GSDIIIa 232400	1p21	***AGL*** *610860*	Amylo-1, 6-glucosidase, 4-alpha-glucanotransferase (glycogen debranching enzyme)	Sheng et al. (1996)	
Glycogen storage disease type IV	9.3	AR	GSDIV 232500	3p12	***GBE1*** *607839*	1,4-α-D=glucan 6-β-D-[1.4-D-glucano] transferase, branching enzyme 1 (glycogen branching enzyme) A	Brown et al. (1966) Bao et al. (1996) Bruno et al. (2004)	
Glycogen storage type V (McArdle)	9.4	AR	232600	11q13	***PYGM*** *608455*	Glycogen phosphorylase, muscle	Mommaerts et al. (1959) Schmidt et al. (1959) Lebo et al. (1984) Tsujino et al. (1993a)	
Glycogen storage type VII (Tarui)	9.5	AR	232800	12q13	***PFKM*** *610681*	Muscle-type phosphofructokinase	Tarui et al. (1965) Nakajima et al. (1991), Howard et al. (1996)	
Glycogen storage disease type IXd (ex type VIII) or muscle phosphorylase kinase defiency	9.6	XR	GSD9D 300559	Xq13	***PHKA1*** *311870*	Phosphorylase b kinase, alpha submit	Wehner et al. (1994) Burwinkel et al. (2004)	
Glycogenosis type XIV	9.7	AR	GSD14 612934	1p31	***PGM1*** *171900*	Phosphoglucomutase 1	Stojkovic et al. (2009)	
Glycogenosis type XV	9.8	AR	GSD15 613507	3q24	***GYG1*** *603942*	Glycogenin 1	Moslemi et al. (2010)	
Glycogen storage disease type 0	9.9	AR	GSD0b 611556	9q13	***GYS1*** *138570*	Glycogen synthase 1	Kolberg et al. (2007)	
Glycogen storage disease of heart, lethal congenital	9.10	AD	261740	7q36	***PRKAG2*** *602743*	Protein kinase, AMP-activated (AMPK)	Burwinkel et al. (2005)	
Polyglucosan storage myopathy	9.11	AR		20p13	***RBCK1*** *610924*	RanBP-type and C3HC4-type zinc finger containing 1 (heme-oxidized IRP2 ubiquitin ligase 1)	Nilsson et al. (2013)	
(B) Glycolytic pathway								
Posphoglycerate kinase deficiencey	9.12	XR	300653	Xq13	***PGK1*** *311800*	Phosphoglycerate kinase	DiMauro et al. (1981a, 1983) Rosa (1982)	

Table (*continued*)

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)
Phosphoglycerate mutase deficiency	9.13	AR	GSD10 261670	7p12-p13	***PGAM2*** *612931*	Phosphoglycerate mutase 2 (muscle)	DiMauro et al. (1981b) Edwards et al. (1989) Castella-Escola et al. (1990) Tsujino et al. (1993b)	
Lactate dehydrogenase-A deficiency	9.14	AR	GSD11 612933	11p15.4	***LDHA*** *150000*	Lactate dehydrogenase A	Kanno et al. (1980) Scrable et al. (1990)	
Enolase deficiency	9.15	AD	GSD13 612932	17pter-p12	***ENO3*** *131370*	Enolase 3, beta, muscle specific	Comi et al. (2001)	
(C) Disorders of lipid metabolism								
Carnitine palmitoyl-transferase deficiency	9.16	AR	255110	1p32	***CPT2*** *600650*	Carnitine palmitoyl transferase II [*M*]	DiMauro et al. (1973) Finocchiaro et al. (1991) Taroni et al. (1993) Gellera et al. (1994)	
Primary systemic carnitine deficiency	9.17	AR	CDSP 212140	5q31	***SLC22A5*** *603377*	Solute carrier family 22, member 5	Nezu et al. (1999)	
Carnitine/acyl-carnitine translocase deficiency	9.18	AR		3p21.31	***SLC25A20*** *212138*	Solute carrier family 25 (carnitine/acylcarnitine translocase), member [*M*]	Huizing et al. (1997), Ogawa et al. (2000)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIA)	9.19	AR	GAIIA 231680	15q23-q25	***ETFA*** *608053*	Electron-transfer-flavoprotein, alpha polypeptide [*M*]	Indo et al. (1991) Freneaux et al. (1992)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIB)	9.20	AR	GAIIB 231680	19q13.3-q13.4	***ETFB*** *130410*	Electron-transfer-flavoprotein, beta polypeptide [*M*]	Colombo et al. (1994)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIC)	9.21	AR	GAIIC 231680	4q32-q35	***ETFDH*** *231675*	Electron-transferring-flavoprotein dehydrogenase [*M*]	Beard et al. (1993)	Allelic to MADD (Group 9)
Acyl-CoA dehydrogenase (very long chain) deficiency (VLCAD deficiency)	9.22	AR	201475	17p13	***ACADVL*** *609575*	Acyl-Coenzyme A dehydrogenase, very long chain [*M*]	Aoyama (1993, 1995) Strauss et al. (1995) Mathur et al. 1999)	
Triglyceride storage disease with ichthyosis [impaired long-chain fatty acid oxidation] (Chanarin-Dorfman syndrome)	9.23	AR	CDS 275630	3p25.3-p24.3	***ABHD5*** *(CGI-58)* *604780*	Abhydrolase domain containing 5	Lefevre et al. (2001)	
Neutral lipid storage disease with myopathy without ichthyosis	9.24	AR	NLSDM 610717	11p15.5	***PNPLA2*** *609059*	Adipose triglyceride lipase = desnutrin	Fischer et al. (2007)	
Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency (Lipid storage myopathy)	9.25	AR	MADD 231680	4q32-q35	***ETFDH*** *231675*	Electrontransferring-flavoprotein dehydrogenase [*M*]	Olsen et al. (2007)	Allelic to GAIIC (group 9)
Recurrent myoglobinuria, autosomal recessive	9.26	AR	268200	2p25.1	***LPIN1*** *605518*	Lipin 1 (phsosphatidic acid phosphatase 1)	Zeharia et al. (2008)	
GROUP 10. HEREDITARY CARDIOMYOPATHIES:								
10-A NON-ARRHYTHMOGENIC CARDIOMYOPATHIES								
(a) Hypertrophic cardiomyopathies								
Familial hypertrophic cardiomyopathy, 1	10.1	AD	CMH1 192600	14q12	***MYH6*** *60710* ***MYH7*** *160760*	Myosin heavy chain 6 (alpha) or 7 (beta), cardiac muscle	Jarcho et al. (1989) Solomon et al. (1990) Tanigawa et al. (1990) Geisterfer-Lowrance et al. (1990)	Allelic to CFTD (group 3), myosin storage myopathy (group 3), MPD1 (group 4), CMD1S (group 10)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Familial hypertrophic cardiomyopathy, 2	10.2	AD	CMH2 115195	1q32	***TNNT2*** *191045*	Cardiac troponin T	Watkins et al. (1993) Thierfelder et al. (1994)	
Familial hypertrophic cardiomyopathy, 3	10.3	AD	CMH3 115196	15q22.1	***TPM1*** *191010*	Tropomyosin-1	Thierfelder et al. (1994)	
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)
Familial hypertrophic cardiomyopathy, 6	10.5	AD	CMH6 600858	7q31	***PRKAG2*** *602743*	Protein kinase, AMP-activated, gamma 2 non-catalytic subunit	Blair et al. (2001)	Allelic to Glycogene storage disease of heart, lethal congenital (group 9)
Familial hypertrophic cardiomyopathy, 7	10.6	AD	CMH7 613690	19q13.4	***TTNI3*** *191044*	Cardiac troponin I	Kimura et al. (1997)	Allelic toRCM1 and CMD2A (group 10)
Familial hypertrophic cardiomyopathy, 8	10.7	AD	CMH8 608751	3p21	***MYL3*** *160790*	Myosin, light chain 3, alkali; ventricular, skeletal, slow	Poetter et al. (1996)	
Familial hypertrophic cardiomyopathy, 9	10.8	AD	CMH9 613765	2q24.3	***TTN*** *188840*	Titin	Satoh et al. (1999)	Allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), HMERF (group 5), CMD1G (group 10)
Familial hypertrophic cardiomyopathy, 10	10.9	AD	CMH10 608758	12q23-q24	***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 1	Mogensen et al. (1999)	Alllelic to CMD1R (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-p14	***TNNC1*** *191040*	Slow troponin C	Landstrom et al. (2008)	Allelic to CMD1Z (group 10)
Familial hypertrophic cardiomyopathy, 14	10.13	AD	CMH14 613251	14q12	***MYH6*** *160710*	Myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al. (2005)	Allelic to CMD1EE (group 10)
Hypertrophic cardiomyopathy with vinculin deficiency,	10.14	AD	CMH15 613255	10q22	***VCL*** *193065*	Vinculin (metavinculin)	Vasile et al. (2006)	Allelic to CMD1U (group 10)
Hypertrophic cardiomyopathy	10.15		CMH 192600	20q13.3	***MYLK2*** *606566*	Myosin light chain kinase 2	Davis et al. (2001)	
Hypertrophic cardiomyopathy	10.16		CMH 192600	3p25	***CAV3*** *601253*	Caveolin-3	Hayashi et al. (2004), Fulizio et al. (2005)	Allelic to LGMD1C (group 1), hyperCKemia (group 5), RMD2 (group 6)
Hypertrophic cardiomyopathy, early-onset fatal by deficit in COX15	10.17	AR		10q24	***COX15*** *603646*	COX15 homolog, cytochrome c oxidase assembly protein [*M*]	Antonicka et al. (2003)	
Hypertrophic cardiomyopathy with myozenin 2 defect	10.18	AD	CMH16 613838	4q26	***MYOZ2*** *605602*	Myozenin 2	Osio et al. (2007)	
Hypertrophic cardiomyopathy	10.19	AD	CMH17 613873	20q13.12	***JPH2*** *605267*	Junctophilin-2	Landstrom et al. (2007) Matsuhita et al. (2007)	
Hypertrophic cardiomyopathy with phospholamban defect	10.20	AD	CMH18 613874	6q22	***PLN*** *172405*	Phospholamban	Minamisawa et al. (2003) Landstrom et al. (2011)	Allelic to dilated cardiomyopathy (group 10)
Hypertrophic cardiomyopathy with nexilin defect	10.21	AD	CMH20 613876	1p31.1	***NEXN*** *613121*	Nexilin (F-actin binding protein)	Wang et al. (2010)	Allelic to dilated CMD1CC (group 10)
Hypertrophic cardiomyopathy with cardiac ankyrin repeat domain protein defect	10.22	AD		10q23.33	***ANKRD1*** *609599*	Ankyrin repeat domain-protein 1 (cardiac)	Arimura et al. (2009)	Allelic to dilated cardiomyopathy (group 19)

Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Hypertrophic cardiomyopathy with actinin-2 defect	10.23	AD		1q43	***ACTN2*** *102573*	actinin alpha2	Chiu C et al. (2010)	Allelic to CMD1AA (group 10)
Hypertrophic mitochondrial cardiomyopathy with NDUFAF1 defect	10.24	AD		15q15.1	***NDUFAF1*** *606934*	NADH-ubiquinone oxidoreductase 1 alpha subcomplex [*M*]	Fassone et al. (2011)	Patient with HCM and isolated respiratory complex I deficiency
Hypertrophic mitochondrial cardiomyopathy with MRPL3 defect	10.25	AR		3q21-q23	***MRPL3*** *607118*	mitochondrial ribosomal protein L3 [*M*]	Galmiche et al. (2011)	
(b) Dilated cardiomyopathies								
Dilated cardiomyopathy, 1A	10.26	AD	CMD1A 115200	1q21	***LMNA*** *150330*	Lamin A/C	Fatkin et al. (1999)	Allelic to EDMD2 (group 1), EDMD3 (group 1); LGMD1B (group 1), CMT2B1 (group 14), [+FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, not in this table]
Dilated cardiomyopathy, 1AA	10.27	AD	CMD1AA 612158	1q42-q43	***ACTN2*** ***102573***	Actinin alpha2	Mohapatra et al. (2003)	
Dilated cardiomyopathy, 1B	10.28	AD	CMD1B 600884	9q13	?		Krajinovic et al. (1995)	
Dilated cardiomyopathy, 1C	10.29	AD	CMD1C 601493	10q22-q23	***LDB3 (ZASP)*** *605906*	LIM domain binding -3 (Z band alternatively spliced PDZ motif)	Bowles et al. (1996), Vatta et al. (2003), Arimura et al. (2004)	Allelic to Markesbery-Griggs (group 4), MFM (group 5)
Dilated cardiomyopathy, 1D	10.30	AD	CMD1D 601494	1q32	***TNNT2*** *191045*	Troponin type 2 (cardiac)	Durand et al. (1995), Kamisago et al. (2000)	
Dilated cardiomyopathy, 1E with conduction disorder and arrythmia	10.31	AD	CMD1E 601154	3p21	***SCN5A*** *600163*	Sodium channel, voltage-gated, type V, alpha	McNair et al. (2004)	Allelic to LQT3 (group 10), Brugada syndrome (group 10), SSS1 (group 10/A)
Dilated cardiomyopathy, 1F	10.32	AD	CMD1F 602067	6q23	?		Messina et al. (1997)	synonymous to LGMD1E (group 1)
Dilated cardiomyopathy, 1G	10.33	AD	CMD1G 604145	2q31	***TTN*** *188840*	Titin	Siu et al. (1999) Gerull et al. (2002) Itoh-Satoh et al. (2002)	Allelic to LGMD2 J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), HMERF (group 5), CMH9 (group 10/A)
Dilated cardiomyopathy, 1H	10.34	AD	CMD1H 604288	2q14-q22	?		Jung et al. (1999)	
Dilated cardiomyopathy, 1I	10.35	AD	CMD1I 604765	2q35	***DES*** *125660*	Desmin	Li et al. (1999)	
Dilated cardiomyopathy, 1J	10.36	AD	CMD1J 605362	6q23-24	***EYA4*** *603550*	Eyes absent homolog	Schönberger et al. (2005a)	
Dilated cardiomyopathy, 1K	10.37	AD	CMD1K 605582	6q12-q16	?		Sylvius et al. (2001)	
Dilated cardiomyopathy, 1L	10.38	AD	CMD1L 606685	5q33	***SGCD*** *601411*	Sarcoglycan, delta	Tsubata et al. (2000)	Allelic to LGMD2F (group 1)
Dilated cardiomyopathy, 1M	10.39	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/A)
Dilated cardiomyopathy, 1N	10.40	AD	CMD1N; 607487	17q12	***TCAP*** *604488*	Telethonin (titin-cap)	Knoll et al. (2002)	Allelic to LGMD2G (group 1)
Dilated cardiomyopathy, 1O	10.41	AD	CMD1O 605569	12p12.1	***ABCC9*** *601439*	ATP-binding cassette, sub-family C (member 9)	Bienengraeber etal (2004)	
Dilated cardiomyopathy, 1P	10.42	AD	CMD1P 609909	6q22	***PLN*** *172405*	Phospholamban	Schmitt et al. (2003), Haghighi et al. (2003, 2006)	Allelic to hypertrophic cardiomyopathy (group 10/A)

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Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Dilated cardiomyopathy, 1Q	10.43	AD	CMD1Q 609915	7q22.3-q31.1	?		Schonberger et al. (2005b)	
Dilated cardiomyopathy, 1R	10.44	AD	CMD1R	15q14	***ACTC1*** *102540*	Actin, alpha, cardiac muscle 1	Olson et al. (1998), Mogensen et al. (1999)	Allelic to CMH (group 10/A)
Dilated cardiomyopathy, 1S	10.45	AD	CMD1S	14q12	***MYH7*** *160760*	Myosin, heavy chain 7, cardiac muscle, beta	Kamisago et al. (2000)	Allelic to Allelic to CFTD (group 3), myosin storage myopathy (group 3), MPD1 (group 4), CMH1 (group 10/A)
Dilated cardiomyopathy, 1T	10.46	AD	CMD1T	12q22	***TMPO*** *188380*	Thymopoietin (lamina-associated polypeptide 2)	Taylor et al. (2005)	
Dilated cardiomyopathy, 1U	10.47	AD	CMD1U 613694	1q42.13	***PSEN2*** *600759*	Presenilin 2	Li et al. (2006)	
Dilated cardiomyopathy, 1W	10.48	AD	CMD1W 611407	10q22	***VCL*** *193065*	Vinculin (metavinculin)	Olson et al. (2002), Vasile et al. (2006)	Allelic to CMH 15 (group 10/A)
Dilated cardiomyopathy, 1Y	10.49	AD	CMD1Y 611878	15q22.2	***TPM1*** *191010*	Alpha-tropomyosin	Olson et al. (2010)	Allelic to CMH3 (group 10/A)
Dilated cardiomyopathy, 1Z	10.50	AD	CMD1Z 611879	3p21-p14	***TNNC1*** *191040*	Slow troponin C	Mogensen et al. (2004)	Allelic to CMH13 (group 10/A)
Dilated cardiomyopathy, related to alpha-crystallin	10.51	AD		11q23.1	***CRYAB*** *123590*	Alpha B crystallin	Inagaki et al. (2006)	Allelic to myofibrillar myopathy (group 5)
Dilated cardiomyopathy related to MYBPC3	10.52	AD	CMD1A 115200	11p11.2	***MYBPC3*** *600958*	Myosin-binding protein C	Hershberger et al. (2010)	Allelic to congenital myopathy and fatal cardiomyopathy (group 3), CMH4 (group 10/A)
Dilated cardiomyopathy related to fukutin	10.53	AR	CMD1X 611615	9q31-q33	***FCMD***	Fukutin	Murakami et al. (2006)	Allelic to LGMD2L (group 1)
Dilated cardiomyopathy related to tafazzin	10.54	XR	CMD3A 300069	Xq28	***TAZ*** *300394*	Tafazzin	Gedeon et al. (1995)	Allelic to BTHS (group 10/A)
Dilated cardiomyopathy related to dystrophin	10.55	XR	CMD3B 302045	Xp21.2	***DMD*** *300377*	Dystrophin	Muntoni et al. (1993) Milasin et al. (1996)	Allelic to DMD (group 1)
Dilated cardiomyopathy related to laminin-alpha-4	10.56	AD		6q21	***LAMA4*** *600133*	Laminin-alpha4	Knöll et al. (2007)	
Dilated cardiomyopathy related to integrin-linked kinase	10.57	AD		11p15.4	***ILK*** *602366*	Integrin-linked kinase	Knöll et al. (2007)	
Dilated cardiomyopathy related to myopalladin	10.58	AD		10q21.1	***MYPN*** *608517*	Myopalladin	Duboscq-Bidot (2008)	
Dilated cardiomyopathy related to ribonucleic acid binding protein	10.59	AD	CMD1DD 613172	10q25.3	***RBM20*** *613171*	RNA binding motif protein 20	Brauch et al. (2009)	
Dilated cardiomyopathy related to cardiac ankyrin repeat protein	10.60	AD		10q23.33	***ANKRD1*** *609599*	Ankyrin repeat domain 1 (cardiac muscle)	Duboscq-Bidot (2009), Moulik et al. (2009)	
Dilated cardiomyopathy related to cardiac troponin I	10.61	AD	CMD2A 611880	19q34	***TNNI3*** *191044*	Cardiac troponin I	Carballo et al. (2009)	Allelic to CMH7 and RCM1
Dilated cardiomyopathy related to alpha-myosin heavy chain	10.62	AD	CMD1EE 613252	14q12	***MYH6*** *160710*	Myosin, heavy chain 6, cardiac muscle, alpha	Carniel et al. (2005)	
Dilated cardiomyopathy related to nexilin	10.63	AD	CMD1CC 613122	1p32-p31	***NEXN*** *613121*	Nexilin(<F-actin binding protein)	Hassel et al. (2009)	

Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Dilated cardiomyopathy related to nesprin-1	10.64	AD		6q25	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Puckelwartz et al. (2010)	Allelic to EDMD with nesprin-1 defect (group 1) SCAR8 (group 13), dilated APC with nesprin-1 defect (group 16)
Dilated cardiomyopathy related to MURC	10.65	AD		9q31.1	***MURC*** *(CAVIN4)*	Muscle-restricted coiled-coil gene	Rodriguez et al. (2011)	
Dilated cardiomyopathy related to DOLK	10.66	AR	CDG1M 610768	9q34.13	***DOLK*** *610746*	Dolichol kinase	Kranz et al. (2007) Lefeber et al. (2011)	
Dilated cardiomyopathy related to GATAD1	10.67	AR	CMD2B 614672	7q21.3	***GATAD1*** *614518*	GATA zinc finger domain containing 1	Theis et al (2011)	
Recessive neonatal isolated DC	10.68	AR	CMD1GG 613642	5p15.33	***SDHA*** *600857*	Succinate dehydrogenase complex, subunit a, flavoprotein [*M*]	Levitas et al (2010)	
(c) Restrictive cardiomyopathies								
Restrictive cardiomyopathy, 1	10.69	AD	RCM1 115210	19q34	***TTNI3*** *191044*	Cardiac troponin I	Mogensen et al. (2003)	Allelic to CMH7 (group 10/A)
Restrictive cardiomyopathy, 2	10.70		RCM2 609578	10	?		Zhang et al. (2005)	
(d) Other non arrhythmogenic hereditary cardiomyopathies								
Pompe disease Glycogenosis, generalized, cardiac form (early and late onset) also listed in group 9	10.71	AR	GSDII 232300	17q25	***GAA*** *606800*	Glucosidase, alpha; acid	Hers (1963), Martiniuk et al. (1990), Wokke et al. (1995)	Allelic to GSDII (group 9)
Cardioskeletal myopathy with neutropenia and abnormal mitochondria (Barth syndrome)	10.72	XR	BTHS 302060	Xq28	***TAZ*** *300394*	Tafazzin	Bolhuis et al. (1991) Bione et al. (1996)	Allelic to CMD3A (group 10/A)
Noncompaction of left ventricular myocardium with congenital heart defects	10.73	AD	LVNC 606617	18q12.1-q12.2	***DTNA*** *601239*	Dystrobrevin, alpha	Ichida et al. (2001)	
Cardiovalvular dysplasia, X-linked (Myxomatous valvular dystrophy)	10.74	XR	XMVD 314400	Xq28	***FLNA*** *300017*	Filamin A, alpha (actin binding protein 280)	Kyndt et al. (1998), Kyndt et al. (2007)	

10-B FOR ARRHYTHMOGENIC CARDIOMYOPATHIES AND RELATED SYNDROMES *See online version of the gene table at http://www.musclegenetable.fr*

GROUP 11. CONGENITAL MYASTHENIC SYNDROMES								
Slow channel syndromes	11.1	AD	SCCMS 601462	2q24-q32	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1 muscle	Sine et al. (1995) Engel et al. (1996b) Croxen et al. (1997)	Allelic to FCCM (group 11)
	11.2	AD	SCCMS 601462	17p11-p12	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1 muscle	Engel et al. (1996b) Gomez et al. (1996)	Allelic to 608931 (group 11)
	11.3	AD	SCCMS 601462	2q33-q34	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Gomez et al. (2002)	Allelic to FCCM (group 11), and 608931 (group 11)
	11.4	AD, AR	SCCMS 601462	17p13	***CHRNE*** ***100725***	Cholinergic receptor, nicotinic, epsilon	Ohno et al. (1995) Gomez et al. (1995) Engel et al. (1996b) Croxen et al. (2002)	Allelic to FCCMS (group 11.7) and 608931 (group 11.10)
Fast channel syndromes	11.5	AR	FCCMS 608930	2q24-q32	***CHRNA1*** *100690*	Cholinergic receptor, nicotinic, alpha 1 muscle	Wang et al. (1999) Shen et al. (2003)	Allelic to SCCMS (group 11)
	11.6	AR	FCCMS 608930	2q33-q34	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Brownlow et al. (2001)	Allelic to SCCMS (group 11), and 608931 (group 11)
	11.7	AR	FCCMS 608930	17p13	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Ohno et al. (1996)	Allelic to SCCMS (group 11) and 608931 (group 11)
Acetylcholine receptor deficiency	11.8	AR	608931	17p11-p12	***CHRNB1*** *100710*	Cholinergic receptor, nicotinic, beta 1 muscle	Quiram et al. (1999)	Allelic to SCCMS (group 11)

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Table (*continued*)

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)
	11.9	AR	608931	2q33-q34	***CHRND*** *100720*	Cholinergic receptor, nicotinic, delta	Shen et al. (2002)	Allelic to SCCMS (group 11), and FCCMS/(group 11)
	11.10	AR	608931	17p13	***CHRNE*** *100725*	Cholinergic receptor, nicotinic, epsilon	Engel et al. (1996a) Ohno et al. (1997)	Allelic to SCCMS (group 11) and FCCMS (group 11)
Congenital myasthenic syndrome related to rapsyn	11.11	AR	608931	11p11	***RAPSN*** *601592*	Rapsyn	Ohno et al. (2002) Ohno et al. (2003) Dunne et al. (2003)	
Congenital myasthenic syndrome related to choline acetyltransferase	11.12	AR	CMS-EA 254210	10q11.2	***CHAT*** *118490*	Choline acetyltransferase	Ohno et al. (2001) Maselli et al. (2003)	
Congenital myasthenic syndrome related end-plate acetylcholinesterase	11.13	AR	EAD 603034	3p24.2	***COLQ*** *603033*	Collagen-like tail subunit (single strand of homotrimer) of asymmetric acetylcholinesterase	Donger et al. (1998) Ohno et al. (1998, 1999, 2000)	
Congenital myasthenic syndrome related to MuSK	11.14	AR	CMS1B 608931	9q31-q32	***MUSK*** *601296*	Muscle-specific receptor tyrosine kinase	Chevessier et al. (2004)	
Familial limb-girdle myasthenia related to DOK7	11.15	AR	CMS1B 254300	4p16.2	***DOK7*** *610285*	Docking protein 7	Beeson et al. (2006), Selcen et al. (2008)	
Familial limb girdle myasthenia related to agrin	11.16	AR	CMS1B 254300	1p36.33	***AGRN*** *103320*	Agrin	Huzé et al. (2009)	
Familial limb girdle myasthenia with tubular aggregates related to GFPT1	11.17	AR	CMSTA1 610542	2p12-p15	***GFPT1*** *138292*	Glutamine-fructose-6-phosphate transaminase 1	Senderek et al. (2011)	
Familial limb-girdle myasthenia with tubular aggregates related to DPAGT1	11.18	AR	CMSTA2 614750	11q23.3	***DPAGT1*** *191350*	Dolichyl-phosphate (UDP-N-acetylglucosamine) N-acetylglucosamine phosphotransferase 1 (GlcNAc-1-P transferase)	Belaya et al. (2012)	
Congenital myasthenic syndrome related to β2-laminin	11.19	AR	608931	3p21	***LAMB2*** *150325*	β2-Laminin	Maselli et al. (2009)	Allelic to Pierson syndrome (congenital nephrosis and ocular defects)
Congenital myasthenic syndrome, type Ia1	11.20	AR	FIM1 605809	17p13	*?*		Christodoulou et al. (1997)	
Sodium-channel myasthenia	11.21	AR	614198	17q23	***SCN4A*** *603967*	Sodium channel, voltage-gated, type IV, alpha	Tsujino et al. (2003)	Allelic to HOKPP2 (group 7), HYPP (group 7), PMC (group 7), K-aggravated myotonia (group 7)
Escobar syndrome (multiple pterygium syndrome)	11.22	AR	EVMPS 265000	2q22-q44	***CHRNG*** *100730*	Cholinergic receptor, nicotinic, gamma	Hoffman et al. (2006) Morgan et al. (2006)	
Myasthenic syndrome, with plectin defect	11.23	AR		8q24-qter	***PLEC1*** *601282*	Plectin	Banwell et al. (1999) Forrest et al. (2010) Selcen et al. (2011)	Allelic to LGMD2Q (group 1) MDEBS (group 5)
Congenital myasthenic syndrome related to ALG2	11.24	AR		9q31.1	***ALG2*** *607905*	Alpha-1,3/1,6-mannosyltransferase	Cossins et al. (2013)	
Congenital myasthenic syndrome related to ALG14	11.25	AR		1p21.3	***ALG14*** *612866*	UDP-N-acetylglucosaminyltransferase subunit	Cossins et al. (2013)	

Table (*continued*)

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 12. MOTOR NEURONE DISEASES								
Spinal muscular atrophy related to SMN1								
Spinal muscular atrophy, type I (Werdnig-Hoffman)	12.1	AR	SMA1 253300	5q11-q13	***SMN1*** *600354*	Survival of motor neuron 1, telomeric	Gilliam et al. (1990) Melki et al. (1990a, 1994) Lefebvre et al. (1995) Bussaglia et al. (1995) Rodrigues et al. (1995) Roy et al. (1995) Hahnen et al. (1997)	Allelic to SMA2 (group 12), SMA3 (group 12), SMA4 (group 12)
Spinal muscular atrophy, type II (intermediate)	12.2	AR	SMA2 253550	5q11-q13	***SMN1*** *600354*	Survival of motor neuron 1, telomeric	Matthijs et al. (1996) Samilchuk (1996)	Allelic to SMA1, SMA3, SMA4 (group 12)
Spinal muscular atrophy, type III (Kugelberg-Welander)	12.3	AR	SMA3 253400	5q11-q13	***SMN1*** *600354*	Survival of motor neuron 1, telomeric	Brzustowicz et al. (1990) Melki et al. (1990b) Lefebvre et al. (1995)	Allelic to SMA1, SMA2, SMA4 (group 12)
Spinal muscular atrophy, type IV, adult form	12.4	AR	SMA4 271150	5q11-q13	***SMN1*** *600354*	Survival of motor neuron 1, telomeric	Brahe et al. (1995) Clermont et al. (1995)	Allelic to SMA1, SMA2, SMA3 (group 12)
Distal spinal muscular atrophy, recessive								
Spinal muscular atrophy, distal autosomal recessive 1 (with respiratory distress)	12.5	AR	DSMA1 (SMARD1) 604320	11q13.2-q13.4	***IGHMBP2*** *600502*	Immunoglobulin mu-binding protein 2	Grohmann et al. (1999, 2001)	
Spinal muscular atrophy, distal autosomal recessive 2	12.6	AR	DSMA2 605726	9p21	*?*		Christodoulou et al. (2000)	
Spinal muscular atrophy, distal autosomal recessive 3	12.7	AR	DSMA3 607088	11q13	***?***		Viollet et al. (2004)	
Spinal muscular atrophy, distal autosomal recessive 3	12.8	AR	DSMA4 611067	1p36	***PLEKHG5*** *611101*	Pleckstrin homology domain containing, family G (with RhoGef domain) member 5	Maystadt et al. (2006; 2007)	
Distal spinal muscular atrophy, dominant								
Neuronopathy, distal hereditary motor, type I	12.9	AD	HMN1 182960	7q34-q36	***?***		Gopinath et al. (2007)	
Neuronopathy, distal hereditary motor, type I	12.10	AD	HMN2A 158590	12q24	***HSPB8*** *608014*	Heat shock protein 8	Timmerman et al. Irobi et al. (2004)	Allelic to CMT2L (group 14)
Neuronopathy, distal hereditary motor, type II, adult juvenile	12.11	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	12.12	AD	HMN2C 613376	5q11.2	***HSPB3*** *604624*	Heat shock 27 kDa protein 3	Kolb et al. (2010)	
Dominant distal hereditary motor neuropathy	12.13	AD	dHMN	16q22.1	***AARS*** 601065	Alanyl-tRNA synthetase	Zhao et al. (2012)	Allelic to CMT2N (group 14) 613287
Distal spinal muscular atrophy, distal with upper limb predominance (type V)	12.14	AD	DSMAV (HMN5) 600794	7p15	***GARS*** *600287*	Glycyl tRNA synthetase	Christodoulou et al. (1995), Antonellis et al. (2003)	Allelic to CMT2D (group 14)
Distal spinal muscular atrophy type V	12.15	AD	DSMAV (HMN5A) 600794	11q13	***BSCL2*** *606158*	Seipin	Windpassinger et al. (2004)	Allelic to SPG17 (group 15)
Distal spinal muscular atrophy, type VB	12.16	AD	DSMAVB (HMNS5B) 614751	2p11.2	***REEP1*** *609139*	Receptor accessory protein 1 [*M*]	Beetz et al. (2012)	SPG 31 (group 15)
Spinal muscular atrophy, distal, with vocal cord paralysis (Harper-Young)	12.17	AD	HMN7A 158580	2q12.31	***SLC5A7 600***8761	Solute carrier family 5 (sodium/choline cotransporter), member 7	McEntagart et al. (2001), Barwick et al. (2012)	

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Table (*continued*)

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)
Distal hereditary motor neuronopathy type VIIB	12.18	AD	HMN7B 607641	2p13	***DCTN1*** *601143*	Dynactin 1	Puls et al. (2003)	Allelic to ALS related to DNCT1 (group 12)
Hereditary motor and sensory neuropathy V	12.19	AD	HMSN5 600361	4q34-q35	?		Muglia et al. (2008)	
Spinal muscular atrophy, distal, X-linked, related to UBA1	12.20	XR	SMAX2 301830	Xp11.23	***UBA1 (=UBE1)*** *314370*	Ubiquitin-activating enzyme 1	Ramser et al. (2013), Diamini et al. (2013)	
Spinal muscular atrophy, distal, X-linked	12.21	XR	SMAX3 300489	Xq13-q21	***ATP7A*** *300011*	ATPase, Cu++ transporting, alpha polypeptide	Takata et al. (2004), Kennerson et al. (2010)	
Spinal muscular atrophy, distal related to DNAJB2	12.22	AR	DSMA5 614881	2q32-q34	***DNAJB2*** (*HSJ1*) *604139*	DnaJ (Hsp40) homolog, subfamily B, member 2	Blumen et al. (2012)	
Spinal muscular atrophy congenital non progressive of lower limbs	12.23	AD	SMAL 600175	12q24.1	***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 10/A) allelic to CMT2C (group 14)
Scapuloperoneal spinal muscular atrophy	12.24	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 SMAL (group 10), CMT2C (group 14)
Spinal muscular atrophy, lower extremity, autosomal dominant	12.25	AD	SMALED 158600	14q32	***DYNC1H1*** *600112*	Dynein, cytoplasmic 1, heavy chain 1	Harms et al. (2010, 2012)	CMT2O
Spinal muscular atrophy, lower extremity, autosomal dominant 2	12.26	AD	SMALED2 615290	9q22.31	***BICD2*** *609797*	Bicaudal D homolog 2 (Drosophila)	Neveling et al. (2013), Oates et al. (2013), Peeters et al. (2013)	
Early-onset spinal muscular atrophy, lower extremity, autosomal dominant	12.27	AD		14 q	*?*		Harms et al. (2010)	
Spinal muscular atrophy with progressive myoclonic epilepsy	12.28	AR	SMAPME 159950	8p22	***ASAH1*** *613468*	N-acylsphingosine amidohydrolase (acid ceramidase) 1	Zhou et al. (2012)	
Spinal muscular atrophy, late-onset, Finkel type	12.29	AD	182980	20q13	***VAPB*** *605704*	Vesicle-associated membrane protein-associated protein B and C	Nishimura et al. (2004a)	See also ALS8 (12.37)
Early-onset spinal muscular atrophy, lower extremity, autosomal dominant	12.30	AD		14q	*?*		Harms et al. (2010)	
Autosomal dominant late-onset spinal motor neuropathy	12.31	AD		22q11.2-q13.2	*?*		Penttilä et al. (2012)	
Amyotrophic lateral sclerosis (ALS)								
Familial myotrophic lateral sclerosis 1 (dominant)	12.32	AD	ALS1 105400	21q22	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Siddique et al. (1991, 1996) Rosen et al. (1993)	
Familial amyotrophic lateral sclerosis 1 (recessive)	12.33	AR	ALS1 105400	21q22	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Andersen et al. (1995)	Allelic to IAHSP (group 15)
Amyotrophic lateral sclerosis 2 juvenile	12.34	AR	ALS2 205100	2q33	***ALS2*** *606352*	Alsin	Hentati et al. (1994a) Yang et al. (2001) Hadano et al. (2001)	
Amyotrophic lateral sclerosis 3	12.35	AR	ALS3 606640	18q21	*?*		Hand et al. (2002)	
Amyotrophic lateral sclerosis 4	12.36	AD	ALS4 602433	9q34	***SETX*** *608465*	Senataxin	Chance et al. (1998) Chen et al. (2004) Moreira et al. (2004)	Allelic to AOA2 (group 13)
Amyotrophic lateral sclerosis 5	12.37	AD	ALS5 602099	15q15-q21	*?*		Hentati et al. (1998)	

Table (*continued*)

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Amyotrophic lateral sclerosis 6	12.38	AD	ALS6 608030	16p11.2	***FUS*** *137070*	Fusion (involved in t(12;16) in malignant liposarcoma)	Sapp et al. (2003) Abalkhail et al. (2003) Kwiatkowski et al. (2009) Vance et al. (2009)	
Amyotrophic lateral sclerosis 7	12.39	AD	ALS7 608031	20p13	*?*		Sapp et al. (2003)	
Amyotrophic lateral sclerosis 8	12.40	AD	ALS8 608627	20q13	***VAPB*** *605704*	Vesicle-associated membrane protein-associated protein B and C	Nishimura et al. (2004a, 2004b)	
Amyotrophic lateral sclerosis 9	12.41	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	12.42	AD	ALS10 612069	1p36.2	***TARDBP*** *605078*	TAR DNA-binding protein	Sreedharan et al. (2008)	
Amyotrophic lateral sclerosis 11	12.43	AD	ALS11 612577	6q21	***FIG4*** (***KIAA0274***) *609390*	Sac domain-containing inositol phosphatase 3	Chow et al. (2009)	Allelic to CMT4J (group 14)
Amyotrophic lateral sclerosis 12	12.44	AD AR	ALS12 613435	10p14	***OPTN*** *602432*	Optineurin	Maruyama et al. (2010)	
Amyotrophic lateral sclerosis 13	12.45	AD	ALS13 183090	12q24.12	***ATXN2*** *601517*	Ataxin 2	Elden et al. (2010) Daoud et al. (2011) Van Damme et al. (2011)	Allelic to spinocelular ataxia (group 13)
Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia	12.46	AD	ALS14 613954	9p13.3	***VCP*** *601023*	Valosin-containing protein	Johnson et al. (2011)	Allelic to distal myopathy with VCP defect (group 4), and IBMFD (group 5)
Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	12.47	XD	ALS15 300857	Xp11.21	***UBQLN2*** *300264*	Ubiquilin 2	Deng et al. (2011)	
Amyotrophic lateral sclerosis 16, juvenile	12.48	AR	ALS16 614373	9p13.3	***SIGMAR1*** *601978*	Sigma non-opioid intracellular receptor 1	Al-Saif et al. (2011)	
Amyotrophic lateral sclerosis 17	12.49	AD	ALS17	3p11.2	***CHMP2B*** *609512*	Charged multivesicular body protein 2B	Parkinson et al. (2006) Cox et al. (2010)	
Amyotrophic lateral sclerosis 18	12.50	AD	ALS18 614808	17p13.2	***PFN1*** *176610*	Profilin 1	Wu et al. (2012)	
Susceptibility to amyotrophic lateral sclerosis related to NEFH	12.51	AD	ALSDC 105400	22q12.2	***NEFH*** *162230*	Neurofilament, heavy polypeptide	Al-Chalabie et al. (1999)	
Susceptibility to amyotrophic lateral sclerosis related to peripherin	12.52	AD	105400	12q13.12	***PRPH*** *170710*	Peripherin	Gros-Louis et al. (2004) Leung et al. (2004)	
Susceptibility to amyotrophic lateral sclerosis related to dynactin 1	12.52	AD	105400	2p13.1	***DCTN1*** *601143*	Dynactin 1	Munch et al. (2005)	Allelic to HMN7B (group 12)
Amyotrophic lateral sclerosis and/or frontotemporal dementia	12.53	AD	ALSFTD 105550	9p21.2	***C9orf72*** *614260*	Chromosome 9 open reading frame 72	Morita et al. (2006) DeJesus-Hernandez (2011)	
Others								
Kennedy disease	12.54	XR	SBMA 313200	Xq13	***AR*** *313700*	Androgen receptor	Fishbeck et al. (1986) La Spada et al. (1991)	
Lethal Congenital Contracture Syndrome 1	12.55	AR	LCCS1 253310	9q34	***GLE1*** *603371*	GLE1 RNA export mediator homolog (yeast)	Makela-Bengs et al. (1998) Nousiainen et al. (2008)	
Lethal Congenital Contracture Syndrome 2	12.56	AR	LCCS2 607598	12q13	***ERBB3*** *190151*	v-erb-b2 erythroblastic leukemia viral oncogene homolog 3 (avian)	Narkis et al. (2007)	
Lethal Congenital Contracture Syndrome 3	12.57	AR	LCCS3 611359	19p13	***PIP5K1C*** *606102*	Phosphatidylinositol-4-phosphate 5-kinase, type I, gamma	Narkis et al. (2007)	

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Table (*continued*)

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Spinal muscular atrophy with pontocerebellar hypoplasia, type 1b	12.58	AR	PCH1B 614678	9p13.2	***EXOSC3*** *606489.*	Exosome component 3	Rudnik-Schöneborn et al. (2013)	
Spinal muscular atrophy with pontocerebellar hypoplasia	12.59	AR	PCH1 607596	14q32	***VRK1*** *602168*	Vaccinia related kinase 1	Renbaum et al. (2009)	
Brown-Vialetto-van Laere syndrome 1	12.60	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	12.61	AR	BVVLS2 614707	8q24	***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	12.63	AD		5q13.3	***HEXB*** *606873*	Hexosaminidase B	Rattay et al. (2013)	Sandhoff disease, adult type (OMIM #268800)

GROUP 13. HEREDITARY ATAXIAS *See online version of the gene table at http://www.musclegenetable.fr*

GROUP 14. HEREDITARY MOTOR SENSORY NEUROPATHIES (HMSN)

(A) Charcot–Marie–Tooth neuropathy, type I (demyelinating)

Autosomal dominant (AD-CMT1)

Type 1A	14.1	AD	CMT1A 118220	17p11.2	***PMP22*** *601097*	Peripheral myelin protein 22	Vance et al. (1989) [add Patel et al. (1992) already in the list] Matsunami et al. (1992), Timmerman et al. (1990, 1992), Valentijn et al. (1992) Roa et al. (1993a)	Allelic toCMT1E (group 14), HNPP (group 14), DSS (group 14)
Type 1B	14.2	AD	CMT1B 118200	1q22	***MPZ*** *159440*	Myelin protein zero	Bird et al. (1982) Guiloff et al. (1982) Hayasaka et al. (1993a) Kulkens et al. (1993)	Allelic to CMT2I (group 14), CMT2 J (group 14), DSS (group 14), CMT4E (group 14)
Type 1C	14.3	AD	CMT1C 601098	16p13	***LITAF*** *603795*	Lipopolysaccharide-induced TNF factor	Street et al. (2002, 2003)	
Type 1D	14.4	AD	CMT1D 607678	10q21.1	***EGR2*** *129010*	Early growth response 2 (Krox-20 homolog)	Warner et al. (1998), Street et al. (2003)	Allelic to CMT4E (group 14), DSS (group 14)
Type 1E (with deafness)	14.5	AD	CMT1E 118300	17p11.2	***PMP22*** *601097*	Peripheral myelin protein 22	Kovach et al. (1999), Boerkoel et al. (2002)	Allelic to CMT1A (group 14), DSS (group 14),
Hereditary Neuropathy with Liability to Pressure Palsies	14.6	AD	HNPP 162500	17p11.2	***PMP22*** *601097*	Peripheral myelin protein P22	Chance et al. (1993) Nicholson et al. (1994) Mariman et al. (1994)	Allelic to CMT1A (group 14) CMT1E (group 14), HNPP (group 14), DSS (group 14)
Type 1F	14.7	AD	CMT1F 607734	8p21	***NEFL*** *162280*	Neurofilament, light polypeptide 68 kDa	Jordanova et al. (2003)	Allelic to CMT2E (group 14)
CMT with Congenital vertical talus	14.8	AD	192950	2q31-q32	***HOXD10 (HOX4)*** *142984*	Homeobox D10	Shrimpton et al. (2004)	
Slowed nerve conduction velocity	14.9	AD	NCV 608236	8p23	***ARHGEF10*** *608136*	Rho guanine-nucleotide exchange factor-10	De Jonghe et al. (1999), Verhoeven et al. (2003)	
CMT related to fibulin	14.10	AD		14q32.12	***FBLN5*** *604580*	Fibulin 5 (extra-cellular matrix)	Auer-Grumbach et al. (2011)	

Dominant intermediate (CMTDI)

Type A	14.11	AD	CMTDIA 606483	10q24.1-q25.1	***?***		Verhoeven et al. (2001)	
Type B	14.12	AD	CMTDIB 606482	19p12-13.2	***DNM2*** *602378*	Dynamin 2	Zuchner et al. (2005)	Allelic to CNM (group 3)
Type C	14.13	AD	CMTDIC 608323	1p35	***YARS*** *603623*	Tyrosyl-tRNA synthetase	Jordanova et al. (2003, 2006)	
Type D	14.14	AD	CMTDID 607791	1q22	***MPZ*** *159440*	Myelin protein zero	Mastaglia et al. (1999)	Allelic to CMT1B, CMT4E, CMT2I, CMT2J, DSS (this group)
Type E with glomerulopathy	14.15	AD	CMTDIE 614455	14q32–33	***INF2*** *610982*	Inverted formin 2	Boyer et al. (2011)	

Table (*continued*)

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)
Type F	14.16	AD	CMTD1F 615185	3q28-q29	***GNB4*** *610863*	Guanine nucleotide-binding protein, beta-4	Soong et al. (2013)	
Autosomal recessive (AR-CMT1 or CMT4)								
CMT, type 4A	14.17	AR	CMT4A (=CMT2H) 214400	8q13-q21	***GDAP1*** *606598*	Ganglioside induced differentiation associated protein1 (connexin 32)	Ben Othmane et al. (1993b), Baxter et al. (2002), Cuesta et al. (2002), Nelis et al. (2002)	Allelic to CMT2K ans Autosomal recessive CMT2C (group 14)
CMT, type 4B1	14.18	AR	CMT4B1 601382	11q22	***MTMR2*** *603557*	Myotubularin-related protein-2	Bolino et al. (1996, 2000), Previtali et al. (2003)	
CMT, type 4B2	14.19	AR	CMT4B2 604563	11p15	***SBF2 (=MTMR13)*** *607697*	SET binding factor 2	Azzedine et al (2003), Senderek et al. (2004)	
CMT, type 4B3	14.20	AR	CMT4B3	22q13.33	***SBF1 (=MTMR5)*** *603560*	SET binding factor 1	Nakhro et al. (2013)	
CMT, type 4C	14.21	AR	CMT4C 601596	5q32	***SH3TC2*** *608206 (ex-KIAA1985)*	SH3 domain and tetratricopeptide repeats 2	LeGuern et al. (1996), Senderek et al. (2003)	
CMT4D (HMSN Lom, with deafness)	14.22	AR	HMNSL 601455	8q24	***NDRG1*** *605262*	Nmyc downstream regulated gene 1	Kalaydjieva et al. (1996, 2000) Hunter et al. (2003)	
CMT, type 4E (congenital hypomyelinating myopathy)	14.23		CMT4E 605253	10q21.1	***EGR2*** *129010*	Early growth response 2 (Krox-20 homolog)	Warner et al. (1998)	Allelic to CMT1D (group 14)
CMT, type 4E (congenital hypomyelinating myopathy)	14.24		CMT4E 605253	1q22	***MPZ*** *159440*	Myelin protein zero	Warner et al. (1996)	Allelic to CMT1B (group 14), CMT2I (group 14), CMT2J (group 14), DSS (group 14)
CMT, type 4F	14.25	AR	CMT4F 145900	19q13	***PRX*** *605725*	Periaxin	Delague et al. (2000) Guilbot et al. (2001)	Allelic to DSSE (group 14)
CMT, type 4G (type Russe)	14.26	AR	CMT4G 605285	10q22	***HK1*** *142600*	Hexokinase 1	Rogers et al. (2000), Thomas et al. (2001), Hantke et al. (2009), Sevilla et al. (2013)	
CMT, type 4H	14.27	AR	CMT4H 609311	12p11.21	***FGD4*** *611104*	Frabin	De Sandre-Giovannoli et al. (2005), Delague et al. (2007) Stendel et al. (2007)	
CMT, type 4J	14.28	AR	CMT4J 611228	6q21	***FIG4 (=KIAA0274)*** *609390*	Sac domain-containing inositol phosphatase 3	Chow et al. (2007)	Allelic to ALS 11 (group 12)
X-linked CMT1								
CMT1, X-linked 1	14.29	XD	CMTX1 302800	Xq13	***GJB1*** *304040*	Gap junction protein, beta 1, 32 kDa (connexin 32)	Bergoffen et al. (1993) Bone et al. (1995)	Allelic to DSS (group 14)
CMT1, X-linked 2	14.30	XR	CMTX2 302801	Xp22.2	?		Ionasecu et al. (1992)	
CMT1, X-linked 3	14.31	XR	CMTX3 302802	Xq26	?		Ionasecu et al. (1992), Huttner et al. (2006)	
CMT1, X-linked 4 (Cowchock syndrome)	14.32	XR	CMTX4 310490	Xq24-q26	***AIFM1*** *300169*	Apoptosis-inducing factor, mitochondrion-associated, 1[M]	Priest et al. (1995), Rinaldi et al. (2012)	
CMT1, X-linked 5 (with hearing loss and optic neuropathy	14.33	XR	CMTX5 311070	Xq22-q24	***PRPS1*** ***311850***	Phosphoribosyl pyrophosphate synthetase 1	Kim et al. (2007)	
CMT1, X-linked 6	14.34	XD	CMTX6 300905	Xp22.11	***PDK3*** *300906*	Pyruvate dehydrogenase kinase, isoenzyme 3[M]	Kennerson et al. (2013)	
Dejerine-Sottas syndrome (DSS or CMT3)								
Déjerine-Sottas hypertrophic neuropathy, dominant	14.35	AD	DSSA 145900	17p11.2	***PMP22*** *601097*	Peripheral myelin protein 22	Roa et al. (1993b)	Allelic to CMT1A (group 14) CMT1E (group 14), HNPP (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.36	AD	DSSB 145900	1q21-q23	***MPZ*** *159440*	Myelin protein zero	Hayasaka et al. (1993b)	Allelic to CMT1B (group 14), CMT2I (group 14), CMT2 J (group 14), CMT4E (group 14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.37	AD (digenic)	DSSC 145900	10q21.Xq13	***EGR2*** *129010 and* ***GJB*** *1304040*	Early growth response 2 (Krox-20 homolog) and gap junction protein, beta 1, 32 kDa (connexin 32)	Chung et al. (2005)	Allelic to CMTX1 (group 14)

(*Continued on next page*)

Table (*continued*)

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)
Déjerine-Sottas hypertrophic neuropathy, recessive	14.38	AR	DSSE (=CMT4F) 145900	19q13	***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								
CMT2 Autosomal dominant								
Type 2A1	14.39	AD	CMT2A1 118210	1p36.2	***KIF1B*** *605995*	Kinesin family member 1B	Zhao et al. (2001a)	
Type 2A2	14.40	AD	CMT2A2 609260	1p36.2	***MFN2*** *608507*	Mitofusin 2 [*M*]	Ben Othmane et al. (1993a) Züchner et al. (2004)	
Type 2B	14.41	AD	CMT2B 600882	3q21	***RAB7*** *602298*	RAB7, member of RAS oncogene family)	Kwon et al. (1995) Pericak-Vance et al. (1997) Kok et al. (2003)	
Type 2C	14.42	AD	CMT2C 606071	12q23-q24	***TRPV4*** *600175*	*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)	
Type 2D	14.43	AD	CMT2D 601472	7p15	***GARS*** *600287*	Glycyl tRNA synthetase	Ionasescu et al. (1996) Antonellis et al. (2003)	Allelic to DSMAV (group 12)
Type 2E	14.44	AD	CMT2E 607684	8p21	***NEFL*** *162280*	Neurofilament, light polypeptide 68 kDa	Birouk et al. (2003), Claramunt et al. (2005)	Allelic to SMAL and SPSMA (group 12)
Type 2F	14.45	AD	CMT2F 606595	7q11-q21	***HSPB1*** *602195*	Heat-shock 27-kD protein-1	Ismailov et al. (2001), Evgrafov et al. (2004)	
Type 2G	14.46	AD	CMT2G 608591	12q12-q13	**?**	?	Nelis et al. (2004)	
Type 2H	14.47	AD	CMT2H 607731	8q21.3	**?**	?	Barhoumi et al. (2001)	Maybe Allelic to CMT4A (group 14)
Type 2I (late onset)	14.48	AD	CMT2I 607677	1q22	***MPZ*** *159440*	Myelin protein zero	Auer-Grumbach et al. (2003)	Allelic to CMT1B, CMT2J, DSS, CMT4E (group 14)
Type 2J (with hearing loss and pupillary abnormality)	14.49	AD	CMT2J 607736	1q22	***MPZ*** *159440*	Myelin protein zero	De Jonghe et al. (1999), Chapon et al. (1999)	Allelic to CMT1B, CMT2J, DSS, CMT4E (group 14)
Type 2K	14.50	AD, AR	CMT2K 607831	8q13-q21	***GDAP1*** *606598*	Ganglioside-induced differentiation-associated protein 1	Nelis et al. (2002), Birouk et al. (2003), Claramunt et al. (2005)	Allelic to CMT4A and ARCMT2C (group 14)
Type 2L	14.51	AD	CMT2L 608673	12q24	***HSPB8*** *608014*	Heat shock protein 8	Tang et al. (2004, 2005)	Allelic to HMN2A (group 12)
Type 2N	14.52	AD	CMT2N 613287	16q22.1	**AARS** *601065*	AARS alanyl-tRNA synthetase	Latour et al. (2010)	
Type 2O	14.53	AD	CMT2O 614228	14q32.31	**DYNC1H1** *600112*	Dynein, cytoplasmic 1, heavy chain 1	Weedon et al. (2011)	
Type 2P	14.54	AD	CMT2P 614436	9q33.3	***LRSAM1*** *610933*	leucine rich repeat and sterile alpha motif containing 1	Guernsey et al. (2010) Weterman et al. (2012) Nicolaou et al. (2012)	
Hereditary motor and sensory neuropathy, Okinawa type	14.55	AD	HMSN0/HMNSP 604484	3q13	***TFG*** *602498*	TRK-fused gene	Takeshima et al. (1997, 1999), Maeda et al. (2007), Ishiura et al. (2012), Lee et al. (2013)	
CMT2 (late-onset)	14.56			12q13.3	***MARS*** *156560.*	methionyl-tRNA synthetase	Gonzalez et al. (2013)	
CMT2 Autosomal recessive								
Autosomal recessive CMT2A	14.57	AR	CMT2B1 605588	1q21.2	***LMNA*** *150330*	Lamin A/C	Bouhouch et al. (1999) De Sandre et al. (2002), Worman and Bonne (2007)	Allelic to EDMD2, EDMD3, LGMD1B (group 1) [+FPLD2/ 151660, HGPS/176670, restrictive dermopathy/ 275210, not in this table]

Table (*continued*)

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)
Autosomal recessive CMT2B	14.58	AR	CMT2B2 605589	19q13	***MED25*** *610697*	Mediator complex subunit 25	Leal et al. (2001, 2009)	Allelic to CMT4A, and CMT2K (group 14)
Axonal neuropathy with myotonia	14.59	AR	NMAN 137200	5q23.3	***HINT1*** *601314*	Histidine triad nucleotide binding protein	Zimon et al. (2012)	
Axonal neuropathy recessive	14.60	AR	CMTRIB 613641	16q23.1	***KARS*** *601421*	Lysyl-tRNA synthetase	McLaughlin et al. (2010)	
Axonal neuropathy intermediate recessive C	14.61	AR	CMTRIC 615376	1p36	***PLEKHG5*** *611101*	Pleckstrin homology domain containing, family G (with RhoGef domain) member 5	Azzedine et al. (2013), Kim et al. (2013)	Allelic to DSMA4 (12.8)
(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 HSMN syndromes								
Hereditary sensory and autonomic neuropathy type I	14.62	AD	HSAN1 162400	9q22.1-q22.3	***SPTLC1*** *605712*	Serine palmitoyltransferase 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	14.63	AD	HSAN1B 608088	3p24-p22	**?**		Kok et al. (2004)	
Hereditary sensory and autonomic neuropathy type I	14.64	AD	HSN IC 613640	14q24.3	***SPTLC2*** 605713	Serine palmitoyltransferase long chain base subunit 2	Rotthier et al. (2010)	
Hereditary sensory neuropathy type I.D	14.65	AD	HSN ID 613708	14q22.1	***ATL1*** 606439	atllastin	Guelly et al.	
Hereditary sensory neuropathy, type IIC	14.66	AD	HSN IC 614213	2q37.3	***KIF1A*** 601255	HSN IIC 614213	Riviere et al. (2011)	Allelic to SPG30 (group 15)
Hereditary sensory and autonomic neuropathy type II	14.67	AR	HSAN2 201300	12p.13	***WNK1*** *605232*	WNK lysine deficient protein kinase 1	Lafreniere et al. (2004), Shekarabi et al. (2008)	
Hereditary sensory and autonomic neuropathy type III (Familial dysautonomia, Riley-Day syndrome)	14.68	AR	HSAN3 223900	9q31	*IKBKAP* *603722*	Inhibitor of kappaB kinase complex associated protein	Blumenfeld et al. (1993), Anderson et al. (2001), Slaugenhaupt et al. (2001)	
Hereditary sensory and autonomic neuropathy type V	14.69	AR	HSAN5 608654	1p13.1	***NGFB*** *162030*	Nerve growth factor (beta polypeptide)	Einarsdottir et al. (2004)	
Hereditary sensory neuropathy with dementia and hearing loss.	14.70	AD	HSN1E 614116	19p13.2	**DNMT1** *126375*	DNA (cytosine-5)-methyltransferase 1	Klein et al. (2011)	
Peripheral neuropathy and agenesis of the corpus callosum (Charlevoix disease)	14.71	AR	ACCPN 218000	15q13-q14	***SLC12A6 (KCC3)*** *604878*	Solute carrier family 12 (potassium chloride cotransporter)	Casaubon et al. (1996) Howard et al. (2002a, 2002b)	
Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy)	14.72	AD	HNA 162100	17q25	***SEPT9*** *604061*	Septin 9	Pellegrino et al. (1996) Kuhlenbaumer et al. (2005)	
Giant axonal neuropathy	14.73	AR	GAN 256850	16q24.1	***GAN1*** *605379*	Gigaxonin	Ben Hamida et al. (1997), Bomont et al. (2000)	
Congenital cataracts, facial dysmorphism and neuropathy	14.74	AR	CCFDN 604168	18p23	***CTDP1*** *604927*	CTD phosphatase subunit 1	Varon et al. (2003)	
Complex motor and sensory axonal neuropathy plus microcephaly and cerebral dysgenesis	14.75	AR		14q32.2	***VRK1*** *602168*	Vaccinia related kinase 1	Gonzaga-Jauregui et al. (2013)	

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Table (*continued*)

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 15. HEREDITARY PARAPLEGIAS *See online version of the gene table at http://www.musclegenetable.fr*								
GROUP 16. OTHER NEUROMUSCULAR DISORDERS								
Torsion dystonia, early onset	16.1	AD	EOTD 128100	9q34	***TOR1A (DYT1)*** *605204*	Torsin A	Ozelius et al. (1997) Ikeuchi et al. (1999)	
Myoclonus-dystonia syndrome	16.2	AD	DYT11 159900	7q21	***SGCE*** *604149*	Epsilon-sarcoglycan	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	***IKBKAP*** *603722*	Inhibitor of kappaB kinase complex associated protein	Blumenfeld et al. (1993), Anderson et al. (2001), Slaugenhaupt et al. (2001)	
Familial amyloid neuropathy	16.4	AD	105210	18q12.1	**TTR** *176300*	Transthyretin (prealbumin)	Costa et al. (1978) Tawara et al. (1983), Saraiva et al. (1995)	
Congenital fibrosis of the extraocular muscles	16.5	AD	CFEOM1 135700	12q12	***KIF21A*** *608283*	Kinesin family member 21a	Engle et al. (1994), Yamada et al. (2003), Tiab et al. (2004)	
Congenital fibrosis of the extraocular muscles	16.6	AD	CFEOM2 2602078	11q13	***PHOX2A (ARIX)*** *602753*	Paired-like aristaless homeobox protein 2a	Wang et al. (1998) Nakano et al. (2001)	
Congenital fibrosis of the extraocular muscles	16.7	AD	CFEOM3 600638	16q24	***TUBB3*** *602661*	Tubulin, beta 3	Doherty et al. (1999), Tischfield et al. (2010)	
Distal arthrogryposis type 1	16.8	AD	DA1 108120	9p13	***TPM2*** *190990*	Tropomyosin 2, b tropomyosin	Sung et al. (2003a)	Allelic to nem4 and cap disease (group 3), DA2B (group 16.14)
Distal arthrogryposis type 2a, freeman-Sheldon syndrome	16.9	AD	DA2A 193700	17p13	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al. (2006)	DA2B (group 16)
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.10	AD	DA2B, 601680	11p15	***TNNI2*** *191043*	Troponin I, fast-twitch skeletal muscle isoform	Sung et al. (2003a), Kimber et al. (2006)	
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.11	AD	DA2B, 601680	11p15	***TNNT3*** *600692*	Troponin T3, fast skeletal	Sung et al. (2003b)	
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.12	AD	DA2B, 601680	17p13	***MYH3*** *160720*	Myosin heavy chain 3, skeletal muscle, embryonic	Toydemir et al. (2006a)	
Distal arthrogryposis type 2b, Sheldon-Hall syndrome	16.13	AD	DA2B, 601680	9p13	***TPM2*** *190990*	Tropomyosin 2 (beta)	Tajsharghi et al. (2007c), Ochala et al. (2007)	Nem4 (group 3) Cap disease (group 3), DA1 (group 16)
Arthrogryposis multiplex congenita with nesprin-1 defect	16.14	AR	AMC	6q25	***SYNE1*** *608441*	Spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Attali et al. (2009)	Allelic to nesprin-related muscular dystrophy (group 1), dilated cardiomyopathy (group 10/A) and SCAR8 (group 13)
Trismus-pseudocamp-todactyly	16.15	AD	608837	17p13	***MYH8*** *160741*	Myosin heavy chain, 8, skeletal muscle, perinatal	Veugelers et al. (2004), Toydemir et al. (2006b)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1	16.16	AD	PEOA1 157640	15q25	***POLG*** *174763*	Polymerase, DNA, gamma [*M*]	Van goethem et al. (2001)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2;	16.17	AD	PEOA2 609283	4q35	***SLC25A4*** (*formerly ANT1*) *103320*	Solute carrier family 25 (mitochondrial carrier; adenine nucleotide translocator), member 4 [*M*]	Kaukonen et al. (2000)	

Table (*continued*)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein (mitochondrial proteins indicated by symbol [*M*])	Key references	Other allelic disease(s) (group in this table)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3	16.18	AD	PEOA3 609286	10q24	***PEO1 (C10ORF2)*** *606075*	Twinkle, T7 gene 4-like protein with intramitochondrial nucleoid localization [*M*]	Suomalinen et al. (1997) Spelbrink et al. (2001)	Allelic to iosca (group 13)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4	16.19	AD	PEOA 4610131	10q24	***POLG*** *2604983*	Polymerase, DNA, gamma-2; POLG accessory subunit; POLGB [*M*]	Longley et al. (2006)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5	16.20	AD	PEOA5 613077	8q23	***RRM2B*** *604712*	Ribonucleotide reductase M2 B [*M*]	Tyynismaa et al. (2009)	Allelic to MTDP8B (group 16)
Mitochondrial DNA depletion myopathy	16.21	AR	MTDPS3 609560	16q22	***TK2*** *188250*	Thymidine kinase, [*M*]	Saada et al. (2001)	
Mitochondrial DNA depletion myopathy, encephalomyo-pathic form	16.22	AR	MTDPS5 612073	13q12-q13	***SUCLA2*** *603921*	Succinate-coA ligase, adp-forming,beta subunit [*M*]	Elpeleg et al. (2005)	
Mitochondrial DNA depletion myopathy	16.23	AR	MTDP8B 612075	8q23	***RRM2B*** *604712*	Ribonucleotide reductase, M2B [*M*]	Bourdon et al. (2007)	
Progressive extranal ophthalmoplegia with optic atrophy, optic atrophy 1 with deafness	16.24	AD	125250	3q28-q29	***OPA1*** *605290*	OPA1 protein, dynamin related gtpase [*M*]	Amati-bonneau et al. (2008) Hudson et al. (2008)	Allelic to PEOA5 (group 16)

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