Neuromuscular Disorders 19 (2009) 77–98

Contents lists available at ScienceDirect

Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd

The 2009 version of the gene table of neuromuscular disorders

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General features

The purpose of this table is to provide the reader of *Neuromuscular Disorders* with an updated list of **monogenic** muscle diseases in which the causative **nuclear** 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.

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; 11. Congenital myasthenic syndromes; 12. Spinal muscular atrophies; 13. Hereditary ataxias; 14. Hereditary motor and sensory neuropathies; 15. *Hereditary paraplegias;* 16. *Other neuromuscular disorders.*

In each category every entry corresponds to a given clinical entity, with nine descriptive features:

Column 1: Name of disease (where there are several synonymous designations the most commonly used by clinicians is preferred).

Column 2: Alphanumeric temporary annual code to designate the item in each disease category, subject to changes in the subsequent printed versions if intercalations/deletions are needed.

Column 3: Mode of inheritance (AD: autosomal dominant; AR: autosomal recessive; XR: X-linked recessive).

Column 4: Generally accepted locus symbol, with corresponding OMIM[1] locus number.

Column 5: Chromosomal localization of the locus.

Column 6: Gene symbol, approved by the HUGO Gene Nomenclature Committee (HGNC) http://www.genenames.org/, followed by the corresponding OMIM gene number.

Column 7: Protein name (most of the time approved by the HGNC).

Column 8: Key references (in general limited to first locus chromosomal assignment; first identification of the gene; major contribution in the gene pathology).

Column 9: Other allelic disease phenotype(s)

Development of the gene table

Since its creation in the first issue of this journal (1991), the table has exploded in size (new morbid genes, new phenotypes, new morbid loci) and in complexity, essentially due to genetic and phenotypic heterogeneity. In addition the lack of congruence between clinical-based and molecular-based classifications has dismantled the classical nosology, notably blurring the limits of the field of neuromuscular disorders. This situation induced us in 2005 to start, in parallel to the rigid annual printed version, an **online gene table database**, where space is not limited and which is easier to manage and consult *(see below).*

Preparation of the 2009 edition of the gene table

The revision and updating of this version was done under the supervision of the following experts: Kate Bushby (*group 1*), Francesco Muntoni (*groups 2 and 10*) Pascale Guicheney (*groups 2 and 10)*, Ana Ferreiro (*group 3*), Bjarne Udd (*group 4*), Anders Olfors (*group 5 and 16)* , Louis Ptáček (*group 7),* Ichizo Nishino (*group 8*), Salvatore Di Mauro *(group 9),* Andy Engel (*group 11*), Judith Melki (*group 12*), Valérie Delague (*group 14*), Christel Depienne (*group 15*), Haluk Topaloglu (*group 15*).

To save space, the Neuromuscular Disorders editorial board approved the removal from the printed version of some items considered to be "less neuromuscular", the removed material being preserved and maintained on the online version of the table. This change in the 2009 printed table involves three groups: *group 10. Hereditary cardiomyopathies* in which only the arrythmogenic and Long QT syndromes are omitted (42 items following item 10.43); *group 13. Hereditary ataxias*, and *group 15. Hereditary paraplegias,* in which **all** the items are omitted *(48 and 40* respectively*).*

I am extremely appreciative of the invaluable help I was given by Jane Miller at all stages of elaboration of this table.

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[1] OMIM = *Online Mendelian Inheritance in Man*, OMIMTM. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD) and National Center for Biotechnology Information, National Library of Medicine (Bethesda, MD). World Wide Web URL: http://ncbi.nlm.nih.gov/sites/entrez?db=OMIM.

doi:10.1016/j.nmd.2008.11.001

References

Only new key references added since the 2008 edition are printed after the table, arranged by disease group. The last printed cumulative alphabetical list comprising all key references published in the previous isssues (1991 to 2007) is in the gene table published in January 2007 issue (Vol 17, No.1, 81–102). The complete list of current references is available on the online gene table ***(http://www.musclegenetable.org/)***

The online gene table

This version is freely available online at the following URL address : ***http://www.musclegenetable.org.*** Started in 2005, it has been devised, maintained and developed by Dalil Hamroun and Christophe Béroud at the Laboratoire de Génétique Moléculaire, Institut Universitaire de Recherche Clinique, 34093 Montpellier Cedex 5, France. It is fed by all the data selected and classified by the curator of the NMD gene table (JCK). It contains the items provided in the printed version plus those mentioned above that no longer appear in it. In the online version the data are cross-referenced and linked to *PubMed* and to major databases related to molecular medicine (*Leiden Muscular Dystrophy, OMIM, NCBI, Genatlas*). It contains several query tools allowing one to perform a variety of interrogations. It is subject to improvements and will be linked to mutation databases. It is clear that the computerized version of the table is now surpassing the printed version which cannot accommodate the ever-increasing volume and complexity of data. The computer team in Montpellier must be applauded for their skill and dedication in incorporating all new developments into the online gene table so magnificently.

Statistics

The 2009 full version available on the online site contains **481 clinical entries** and **243 distinct identified genes.** These figures reflect altogether the redundancy of clinical entries (a given gene may produce a large number of « diseases », such as in laminopathies), and of the genes listed (a given « disease » may be caused by mutations affecting one among several possible genes, such as in CMT). There are **96 loci** awaiting gene identification (indicated by a question mark in columns 6 and 7 of the printed table).

NOTE

Users of the gene table are kindly requested to send any comment**s** about the printed and/or the online version to *jean-claude.kaplan@inserm.fr*

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Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd

Gene table of monogenic neuromuscular disorders (nuclear genome only)

Vol. 19 No. 1, January 2009

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

DISEASE NAME	Item line in this group	Inheri-tance	Locus or disease symbol and OMIM number	Chromo-some	Gene symbol and OMIM number	Protein	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 (group10)
Emery-Dreifuss muscular dystrophy, X-linked, type 1	1.2	XR	EDMD 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		Xq26.3	***FHL1*** *300163*	four and a half LIM domain 1	Gueneau et al. (2008)	allelic to reducing body myopathy (group 3), XMPMA (group 5), XPMD (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 (group1), CMD1A (group10), CMT2B1 (group14), *[+ 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 604929	1q21.2	***LMNA*** *150330*	lamin A/C	Raffaele di Barletta et al. (2000) Worman and Bonne (2007)	allelic to EDMD2 (group1), LGMD1B (group1), CMD1A (group10), CMT2B1 (group14), *[+ several other phenotypes not in this table: FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, MADA/ 248370]*
Emery-Dreifuss with nesprin-1 defect	1.6	AD		6q25	***SYNE1*** *608441*	spectrin repeat containing, nuclear envelope 1 (nesprin-1)	Zhang et al. (2007)	allelic to SCAR8 (group 13)
Emery-Dreifuss with nesprin-2 defect	1.7	AD		4q23	***SYNE2*** *608442*	spectrin repeat containing, nuclear envelope 2 (nesprin-2)	Zhang et al. (2007)	

(*continued on next page*)

DISEASE NAME	Item line in this group	Inheri-tance	Locus or disease symbol and OMIM number	Chromo-some	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Facio-scapulo-humeral muscular dystrophy	1.8	AD	FSHD 158900	4q35		?	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)	
Limb girdle muscular dystrophies, dominant								
LGMD1A	1.9	AD	LGMD1A 159000	5q31	***MYOT (TTID)*** *609200*	myotilin (titin immunoglobulin domain protein)	Speer et al. (1992), Hauser et al. (2000)	allelic to distal myotilinopathy (group 4), MFM (group 5), sphe roid body myopathy (group 5)
LGMD1B	1.10	AD	LGMD1B 159001	1q11-q21	***LMNA*** *150330*	lamin A/C	van der Kooi et al. (1997) Muchir et al. (2000) Worman and Bonne (2007)	allelic to EDMD2 (group1), EDMD3 (group1), CMD1A (group10), CMT2B1 (group14), [+ several other phenotypes not in this table: FPLD2/ 151660, HGPS/176670, restrictive dermopathy/ 275210, MADA/248370]
LGMD1C	1.11	AD	LGMD1C 607780	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 (group10), LQT9 (group10)
LGMD1D	1.12	AD	LGMD1D 603511	7q	?	?	Speer et al. (1999)	
LGMD1E	1.13	AD	LGMD1E 602067	6q23	?	?	Messina et al. (1997)	Synonymous to CMD1F (group10)
LGMD1F	1.14	AD	LGMD1F 608423	7q32	?	?	Palenzuela et al. (2003)	
LGMD1G	1.15	AD	LGMD1G 609115	4p21	?	?	Starling et al. (2005)	
Limb girdle muscular dystrophies, recessive								
LGMD2A	1.16	AR	LGMD2A 253600	15q15.1	***CAPN3*** *114240*	calpain-3	Beckmann et al. (1991) Young et al. (1992) Richard et al. (1995, 1997)	
LGMD2B	1.17	AR	LGMD2B 253601	2p13	***DYSF*** *603009*	dysferlin	Bashir et al. (1994) Bashir et al. (1998) Liu et al. (1998)	allelic to MM (group 4)
LGMD2C	1.18	AR	LGMD2C 253700	13q12	***SGCG*** *608896*	γ-sarcoglycan	Ben Othmane et al. (1992) Azibi et al. (1993) Noguchi et al. (1995) McNally et al. (1996) Piccolo et al. (1996)	
LGMD2D	1.19	AR	LGMD2D 608099	17q12-q21.33	***SGCA*** *600119*	α-sarcoglycan	Roberds et al. (1994) Piccolo et al. (1995) Passos-Bueno et al. (1995) Ljunggren et al. (1995) Carrié et al. (1997)	
LGMD2E	1.20	AR	LGMD2E 604286	4q12	***SGCB*** *600900*	β-sarcoglycan	Lim et al. (1995) Bönnemann et al. (1995) Bönnemann et al. (1996)	
LGMD2F	1.21	AR	LGMD2F 601287	5q33	***SGCD*** *601287*	δ-sarcoglycan	Passos-Bueno et al. (1996) Nigro et al. (1996)	allelic to CMD1L (group 10)
LGMD2G	1.22	AR	LGMD2G 601954	17q12	***TCAP*** *604488*	titin-cap (telethonin)	Moreira et al. (1997) Moreira et al. (2000)	allelic to CMD1N (group10)
LGMD2H	1.23	AR	LGMD2H 254110	9q31-q34	***TRIM32*** *602290*	tripartite motif-containing 32 (ubiquitin ligase)	Weiler et al. (1998) Frosk et al. (2002)	allelic to sarcotubular myopathy (group 3)
LGMD2I	1.24	AR	LGMD2I 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	1.25	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 (group10), CMD1G (group10)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
LGMD2K	1.26	AR	LGMD2K 609308	9q34	***POMT1*** *607423*	protein-*O*-mannosyl-transferase 1	Balci et al. (2005) D'Amico et al. (2006)	allelic to WWS (group 2)
LGMD2L	1.27	AR	LGMD2L 611307	11p14.3	***ANO5 (TMEM16E)*** *608662*	anoctamin 5	Jarry et al. (2007) Bolduc et al. (2008)	
LGMD2M	1.28	AR	LGMD2M 611588	9q31	***FKTN*** *607440*	fukutin	Murakami et al. (2006) Godfrey et al. (2006)	allelic to FCMD (group 2) WWS (group2) dilated cardiomyopathy (group10)
LGMD2N	1.29	AR	LGMD2N	14q24	***POMT2*** *607439*	protein *O*-mannosyl transferase 2	Biancheri et al. (2007)	allelic to WWS (group 2) and to MEB (group 2)
LGMD2O	1.30	AR	LGMD2O	1p34	***POMGNT1*** *606822*	protein *O*-linked mannose beta1,2-*N*-acetylglucos-aminyltrans-ferase 1	Godfrey et al. (2007) Clement et al. (2008)	allelic to WWS (group 2) and to MEB (group 2)

See also group 4 (Distal myopathies) and group 5 (Other myopathies)

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)	
Muscular dystrophy, congenital	2.2	AR	MDC1B 604801	1q42	*?*	?	Brockington et al. (2000)	
Congenital muscular dystrophy and abnormal glycosylation of dystroglycan	2.3	AR	MDC1C 606612	19q13	***FKRP*** *606596*	fukutin related protein	Brockington et al. (2001b) Topaloglu et al. (2003)	allelic to LGMD2I (group 1), WWS (group 2), MEB (group 2)
Congenital muscular dystrophy and abnormal glycosylation of dystroglycan	2.4	AR	MDC1D 608840	22q12	***LARGE*** *603590*	like-glycosyl transferase	Longman et al. (2003)	
Fukuyama congenital muscular dystrophy	2.5	AR	FCMD 253800	9q31-q33	***FCMD*** *607440*	fukutin	Toda et al. (1993) Kobayashi et al. (1998)	allelic to WWS (group 2)
Walker-Warburg syndrome	2.6	AR	236670	9q31-q33	***FCMD*** *607440*	fukutin	Beltran-Valero de Bernabe (2003)	allelic to LGMD2L (group 1) and Fukuyama (group 2)
Walker-Warburg syndrome	2.7	AR	WWS 236670	9q34	***POMT1*** *607423*	protein-*O*-mannosyl-transferase 1	Beltran-Valero De Bernabe et al. (2002) van Reeuwijk et al. (2006)	allelic to LGMD2K (group 1)
Walker-Warburg syndrome	2.8	AR	WWS 236670	14q24.3	***POMT2*** *607439*	protein-*O*-mannosyl transferase 2	van Reeuwijk et al. (2005)	allelic to LGMD2N (group 1) and MEB (group 2)
Walker-Warburg syndrome	2.9		WWS 236670	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	2.10	AR	WWS 236670	1p3	***POMGNT1*** *606822*	*O*-mannose beta1,2-*N*-acetyl-glucosaminyl transferase	Taniguchi et al. (2003)	allelic to MEB (group 2)
Walker-Warburg Syndrome	2.10a	AR	WWS 236670	22q12	***LARGE*** 603590	Like-glycosyl transferase	Van Reeuwijk et al (2007)	allelic to MDC 1D (group 2)
Muscle–eye–brain disease	2.11	AR	MEB 253280	1p3	***POMGNT1*** *606822*	*O*-mannose beta1,2-*N*-acetyl-glucosaminyl transferase	Yoshida et al. (2001) Taniguchi et al. (2003)	allelic to WWS (group 2)
Muscle–eye–brain disease	2.12	AR	MEB 253280	19q13	***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	2.13	AR	MEB 253280	14q24.3	***POMT2*** *607439*	protein-*O*-mannosyl transferase 2	Mercuri et al. (2006)	allelic to WWS (group 2)

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Rigid spine syndrome	2.14	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 desmin-related myopathy with Mallory bodies (group 5)
Ullrich syndrome	2.15	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	2.16	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)
Ullrich syndrome	2.17	AR	UCMD 254090	2q37	***COL6A3*** *120250*	collagen type VI subunit alpha 3	Demir et al. (2002)	allelic to Bethlem myopathy (group 2)
Bethlem myopathy	2.18	AD	158810	21q22.3	***COL6A1*** *120220*	collagen type VI subunit alpha 1	Jöbsis et al. (1996)	allelic to UCMD (group 2)
Bethlem myopathy	2.19	AD	158810	21q22.3	***COL6A2*** *120240*	collagen type VI subunit alpha 2	Jöbsis et al. (1996)	allelic to UCMD (group 2)
Bethlem myopathy	2.20	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)
Congenital muscular dystrophy with integrin defect	2.21	AR		12q13	***ITGA7*** *600536*	integrin á7	Hayashi et al. (1998)	
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)	
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)	
Nemaline myopathy	3.5	AR	NEM5 605355	19q13	***TNNT1*** *191041*	troponin T type 1 (skeletal, slow)	Johnston et al. (2000)	
Nemaline myopathy	3.6	AD	NEM6 609273	15q	*?*	?	Gommans et al. (2003)	
Nemaline myopathy	3.7	AR	NEM7 610687	14q12	***CFL2*** *601443*	*cofilin 2* (*muscle*)	Agrawal et al. (2007)	
Myopathy, congenital, with fiber-type disproportion	3.8	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	3.9	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)
Myotubular myopathy	3.10	XR	MTM1 310400	Xq28	***MTM1*** *300415*	myotubularin 1	Thomas et al. (1987) Laporte (1996, 1997, 2000)	
Centronuclear myopathy, dominant	3.11	AD	CNM 160150	19.p13.2	***DNM2*** *602378*	dynamin 2	Bitoun et al. (2005)	allelic to DI-CMTB (group 14)
Centronuclear myopathy, recessive	3.12	AR		2q14	***BIN1*** *601248*	amphiphysin	Nicot et al. (2007)	
Central core disease, dominant	3.13	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 ophtalmoplegia (group 3), MHS1 (group 8)
Central core disease, recessive (transient multiminicore myopathy)	3.14	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 ophtalmoplegia (group 3), MHS1 (group 8)
Multiminicore disease with external ophtalmoplegia	3.15	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	3.16	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)
Hyaline body myopathy	3.17	AR	255160	3p22.2-p21.32	*?*	?	Onengut et al. (2004)	

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Hyaline body myopathy, dominant (myosin storage myopathy)	3.18	AD	608358	14q12	***MYH7*** *160760*	myosin, heavy chain 7, cardiac muscle, beta	Tajsharghi et al. (2003) Bohlega et al. (2004) Laing et al. (2005)	allelic to MPD1 (group 4), CMH1 (group 10), CMD1S (group 10)
Myosin storage myopathy and cardiomyopathy, recessive	3.19	AR		14q12	***MYH7*** *160760*	myosin, heavy chain 7, cardiac muscle, beta	Tajsharghi et al. (2007a)	allelic to MPD1 (group 4), CMH1 (group 10),
Cap disease	3.20	AD		9p13	***TPM2*** *190990*	tropomyosin 2, beta	Tajsharghi et al. (2007b) Lehtokari et al. (2007)	allelic to NEM4 (group 3) DA1 (group 16) and DA2B (group 16)
Congenital neuromuscular disease with uniform type 1 fiber	3.21	AD		19q13.1	***RYR1*** *180901*	ryanodine receptor I	Sato et al. (2008)	allelic to CDD (group 3), multi-minicore disease (group 3), MHS1 (group 8)
Congenital myopathy with fatal cardiomyopathy	3.22	AR		2q31	***TTN*** *188840*	titin	Carmignac et al. (2007)	allelic to LGMD2J (group 1), TMD (group 4), HMERF (group 5), CMH9 (group 10), CMD1G (group 10)
Congenital skeletal myopathy and fatal cardiomyopathy	3.23	AR		11p11.2	***MYBPC3*** *600958*	cardiac myosin binding protein-C	Tajsharghi et al. (2008)	allelic to CMH4 (group 10)
Congenital lethal myopathy	3.24	AR		12q11-q12	***CNTN1*** *600016*	contactin-1	Compton et al. (2008)	
Sarcotubular myopathy	3.25	AR		9q31	**TRIM32** *602290*	tripartite motif containing 32 (ubiquitin ligase)	Schoser et al. (2005)	allelic to LGMD2H (group 1)
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), CMD1G (group 10)
Distal myopathy with rimmed vacuoles (Nonaka)	4.3	AR	NM 605820	9p12-p11	***GNE*** *603824*	glucosamine (UDP-*N*-acetyl)-2-epimerase/*N*-acetylmannosamine kinase	Mitrani-Rosenbaum et al. (1996) Ikeuchi et al. (1997) Eisenberg et al. (2001)	
also known as								
Hereditary inclusion body myopathy	4.3a	AR	IBM2 600737	9p12–p11	***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 Myosin storage myopathy (group 3), CMH1 (group 10), CMD1S (group 10)
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	*?*	?	Ahlberg et al. (1999)	
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)
Late onset distal myopathy (Markesbery-Griggs)	4.12	AD		10q22	***LDB3*** (= *ZASP*) *605906*	LIM domain binding-3 (Z band alternatively spliced PDZ motif)	Griggs et al. (2007)	allelic to MFM (group 5)
Dynamin2 related distal myopathy	4.13	AD	CNM 160150	19.p13.2	***DNM2*** *602378*	dynamin 2	Fischer et al. (2006)	allelic to CNM (group 3) and CMTDIB (group 14)

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DISEASE NAME	Item line in this group	Inheri-tance	Locus or disease symbol and OMIM number	Chromo-some	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
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 = ZASP*** *605906*	LIM domain binding-3 (Z band alternatively spliced PDZ motif)	Selcen and Engel (2005) [Wendy: please add this ref to the list]	allelic to Markesbery-Griggs (group 4) and to CMD1C (group 10)
Myofibrillar myopathy with arrhythmogenic right ventricular cardiomyopathy	5.5	AD	MFM/ARVC	10q22	?		Melberg et al. (1999)	
Myofibrillar myopathy, myotilin related	5.6	AD	*MFM 609200*	5q31	***MYOT*** *(=TTID) 604103*	myotilin (titin immunoglobulin domain protein)	Selcen and Engel (2004) Wendy: please add this ref to the list]	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) [Wendy = please add ref Foroud in list]	allelic to LGMD1A (group 1), MFM (group 5)
Myofibrillar myopathy, filamin-C related	5.8	AD	*MYFM 609524*	7q32	***FLNC*** *102565*	filamin C, gamma (actin binding protein 280)	Vorgerd et al. (2005)	
Myofibrillar myopathy with BAG3 defect	5.9	AD		10q25-q26	***BAG3*** *603883*	BCL2-associated athanogene 3	Selcen et al. (2008)	
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	***"VMA21"*** *(provisional symbol for mutated gene at LOC203547 (UniGene Hs 58633)*		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), CMD1G (group 10)
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)	
Muscle hypertrophy	5.15	AR		2q32	***MSTN (=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)
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)

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Episodic muscle weakness, X-linked	5.20	XR	*EMWX 3002211*	Xp22.3	*?*	?	Ryan et al. (1999)	
GROUP 6. MYOTONIC SYNDROMES								
Myotonic dystrophy (Steinert)	6.1	AD	DM1 160900	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 (group 7)*					
Myotonia, recessive (Becker)	6.4	AR	*see under Ion channel muscle diseases (group 7)*					
Rippling muscle disease, dominant	6.5	AD	RMD1 600332	1q41	*?*		Stephan et al. (1994)	
Rippling muscle disease, dominant	6.6	AD	RMD2 606072	3p25	***CAV3*** *601253*	caveolin-3	Betz et al. (2001)	allelic to LGMD1C (group 1), hyper-CKemia (group 5), RMD2 (group 6), CMH (group 10)
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)
Schwartz-Jampel syndrome	6.8	AR	SJS1 255800	1p34-p36.1	***HSPG*** *2142461 (perlecan)*	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), Potassium 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), Potassium 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), Potassium 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) Ptáček et al. (****)	allelic to HYPP (group 7), HOKPP2 (group 7), PMC (group 7)

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Long QT syndromes	7.7		*see under hereditary cardiomyopathies (group 10)*					
(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	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)	
Periodic paralysis, potassium sensitive cardiodysrythmic (Andersen's syndrome)	7.13		*see LQ7 under hereditary cardiomyopathies (group 10)*					

GROUP 8. MALIGNANT HYPERTHERMIAS

Malignant hyperthermia	8.1	AD	MHS1 180901	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), CCD (group 3), minicore myopathy with external ophtalmoplegia (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)
Glycogen storage disease type IIIa	9.2	AR	GSDIIIa 232400	1p21	***AGL*** 610860	amylo-1, 6-glucosidase, 4-alpha-glucano-transferase (glycogen debranching enzyme)	Shen et al. (1996)	

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Glycogen storage disease type IV	9.3	AR	GSDIV 232500	3p12	***GBE1*** 607839	glucan (1,4-alpha-), branching enzyme 1 (glycogen branching enzyme) A	Brown et al. (1966) Bao et al. (1996)	
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 phos-phofructokinase	Tarui et al. (1965) Nakajima et al. (1991) Howard et al. (1996)	
Phosphoglycerate kinase deficiencey	9.6	XR	300653	Xq13	***PGK1*** *311800*	phosphoglycerate kinase	DiMauro et al. (1981a, 1983) Rosa (1982)	
Phosphoglycerate mutase deficiency	9.7	AR		7p12-p13	***PGAM2*** *261670*	phosphoglycerate mutase 2 (muscle)	DiMauro et al. (1981b) Edwards et al. (1989) Castella-Escola et al. (1990) Tsujino et al. (1993b) Edwards et al. (1989) Castella-Escola et al. (1990) Tsujino et al. (1993b)	
(B) Glycolytic pathway								
Lactate dehydrogenase-A deficiency	9.8	AR		11p15.4	***LDHA*** *150000*	lactate dehydrogenase A	Kanno et al. (1980) Scrable et al. (1990)	
Enolase deficiency	9.9	AD		17pter-p12	***ENO3*** *131370*	enolase 3, beta, muscle specific	Comi et al. (2001)	
(C) Disorders of lipid metabolism								
Carnitine palmitoyl-transferase deficiency	9.10	AR	255110	1p32	***CPT2*** *600650*	carnitine palmitoyl transferase II	DiMauro et al. (1973) Finocchiaro et al. (1991) Taroni et al. (1993) Gellera et al. (1994)	
Primary systemic carnitine deficiency	9.11	AR	CDSP 212140	5q31	***SLC22A5*** *603377*	solute carrier family 22, member 5	Nezu et al. (1999)	
Carnitine/acyl-carnitine translocase deficiency	9.12	AR		3p21.31	***SLC25A20*** *212138*	solute carrier family 25 (carnitine/ acylcarnitine translocase), member	Huizing et al. (1997) Ogawa et al. (2000)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIA)	9.13	AR	GAIIA 231680	15q23-q25	***ETFA*** *608053*	electron-transfer-flavoprotein, alpha polypeptide	Indo et al. (1991) Freneaux et al. (1992)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIB)	9.14	AR	GAIIB 231680	19q13.3-q13.4	***ETFB*** *130410*	electron-transfer-flavoprotein, beta polypeptide	Colombo et al. (1994)	
Multiple acyl-CoA dehydrogenase deficiency (MADD; Glutaric aciduria type IIC)	9.15	AR	GAIIC 231680	4q32-q35	***ETFDH*** *231675*	electron-transferring-flavoprotein dehydrogenase	Beard et al. (1993)	allelic to Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency (Group 9)
Acyl-CoA dehydrogenase (very long chain) deficiency (VLCAD deficiency)	9.16	AR	201475	17p13	***ACADVL*** *609575*	acyl-Coenzyme A dehydrogenase, very long chain	Aoyama (1993, 1995) Strauss et al. (1995) Mathur et al. 1999) [Wendy please add these ref to the list]	
Triglyceride storage disease with impaired long-chain fatty acid oxidation (Chanarin-Dorfman syndrome)	9.17	AR	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	9.18	AR	NLSDM 610717	11p15.5	***PNPLA2*** *609059*	adipose triglyceride lipase = desnutrin	Fischer et al. (2007)	

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Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency (Lipid storage myopathy)	9.19	AR		4q32-q35	***ETFDH*** ***231675***	Electron-transferring-flavoprotein dehydrogenase	Olsen et al. (2007)	allelic to MADD (group 9)
Glycogen storage disease type 0	9.20	AR	GSD0b 611556	9q13	***GYS1*** *138570*	Glycogen synthase 1	Kolberg et al. (2007)	
GROUP 10. HEREDITARY CARDIOMYOPATHIES								
(A) Hypertrophic								
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 myosin storage myopathy (group 3), MPD1 (group 4), CMD1S (group 10)
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)	
Familial hypertrophic cardiomyopathy, 7	10.6	AD	CMH7	19q13.4	***TTNI3*** *191044*	cardiac troponin I	Kimura et al. (1997)	allelic to RCM1 (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	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	***MYLZ*** *160781*	myosin, light chain 2, regulatory, cardiac, slow	Poetter et al. (1996)	
Familial hypertrophic cardiomyopathy, 11	10.10	AD	CMH 192600	15q14	***ACTC1*** *102540*	actin, alpha, cardiac muscle 1	Mogensen et al. (1999)	alllelic to CMD1R (group 10)
Familial hypertrophic cardiomyopathy, 12	10.11	AD	CMH 192600	20q13.3	***MYLK2*** *606566*	myosin light chain kinase 2	Davis et al. (2001)	
Familial hypertrophic cardiomyopathy, 13	10.12	AD	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 with vinculin deficiency,	10.13		CMH 192600	10q22	***VCL*** *193065*	vinculin (metavinculin)	Vasile et al. (2006)	allelic to CMD1U (group 10)
(B) Dilated								
Dilated cardiomyopathy, 1A	10.14	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, MADA/248370 not in this table]*
1A A	10.15	AD	CMD1AA 612158	1q42-q43	*ACTN2* *102573*	actinin alpha2	Mohapatra et al. (2003)	
1B	10.16	AD	CMD1B 600884	9q13	*?*	?	Krajinovic et al. (1995)	
1C	10.17	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)
1D	10.18	AD	CMD1D 601494	1q32	***TNNT2*** *191045*	troponin type 2 (cardiac)	Durand et al. (1995) Kamisago et al. (2000) [Wendy: please: add ref Durand et al. (1995) in the list]	

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
1E (with conduction disorder and arrythmia)	10.19	AD	CMD1E 601154	3p21	***SCN5A*** *600163*	sodium channel, voltage-gated, type V, alpha	Olson et al. (1996); McNair et al. (2004)	allelic to LQT3 (group 10), Brugada syndrome (group 10), SSS1 (group 10)
1F	10.20	AD	CMD1F 602067	6q23	*?*	?	Messina et al. (1997)	synonymous to LGMD1E (group 1)
1G	10.21	AD	CMD1G 604145	2q31	***TTN*** *188840*	titin	Siu et al. (1999) Gerull et al. (2002) Itoh-Satoh et al. (2002)	allelic to LGMD2J (group 1), congenital myopathy with fatal cardiomyopathy (group 3), TMD (group 4), HMERF (group 5), CMH9 (group 10),
1H	10.22	AD	CMD1H *604288*	2q14-q22	*?*	?	Jung et al. (1999)	
1I	10.23	AD	CMD1I 604765	2q35	***DES*** *125660*	desmin	Li et al. (1999)	
1J	10.24	AD	CMD1J 605362	6q23-24	***EYA4*** *603550*	Eyes absent homolog	Schönberger et al. (2005a)	
1K	10.25	AD	CMD1K 605582	6q12-q16	*?*	?	Sylvius et al. (2001)	
1L	10.26	AD	CMD1L 606685	5q33	***SGCD*** *601411*	sarcoglycan, delta	Tsubata et al. (2000)	allelic to LGMD2F (group 1)
1M	10.27	AD	CMD1M 607482	11p15.1	***CSRP3*** *600824*	Cysteine and glycine-rich protein 3 (cardiac LIM protein)	Knoll et al. (2002)	
1N	10.28	AD	CMD1N; 607487	17q12	***TCAP*** *604488*	Telethonin (titin-cap)	Knoll et al. (2002)	allelic to LGMD2G (group 1)
1O	10.29	AD	CMD1O 605569	12p12.1	***ABCC9*** *601439*	ATP-binding cassette, sub-family C (member 9)	Bienengraeber etal (2004)	
1P	10.30	AR	CMD1P 609909	6q22	***PLN*** *172405*	phospholamban	Schmitt et al. (2003) Haghighi et al. (2003, 2006)	
1Q	10.31	AD	CMD1Q 609915	7q22.3-q31.1	*?*	?	Schonberger et al. (2005b)	
1R	10.32	AD	CMD1R	15q14	***ACTC1*** *102540*	actin, alpha, cardiac muscle 1	Olson et al. (1998) Mogensen et al. (1999)	allelic to CMH (group 10)
1S	10.33	AD	CMD1S	14q12	***MYH7*** *160760*	myosin, heavy chain 7, cardiac muscle, beta	Kamisago et al. (2000)	allelic to myosin storage myopathy (group 3), MPD1 (group 4), CMH1 (group 10)
1T	10.34	AD	CMD1T	12q22	***TMPO*** *188380*	Thymopoietin (lamina-associated polypeptide 2)	Taylor et al. (2005)	
1U	10.35	AD	CMD1W 611407	10q22	***VCL*** *193065*	vinculin (metavinculin)	Olson et al. (2002) Vasile et al. (2006)	allelic to CMH (group 10)
1Z	10.36	AD	CMD1Z 611879	3p21-p14	***TNNC1*** *191040*	slow troponin C	Mogensen et al. (2004)	
Dilated cardiomyopathy due to fukutin defect	10.37	AR		9q31-q33	***FKTN*** *607440*	fukutin	Murakami et al. (2006)	allelic to LGMD2M (group 1)
Dilated cardiomyopathy due to tafazzin defect	10.38	XR	CMD3A 300069	Xq28	***TAZ*** *300394*	tafazzin	Gedeon et al. (1995)	allelic to BTHS (group 10)
Dilated cardiomyopathy due to dystrophin defect	10.39	XR	CMD3B 302045	Xp21.2	***DMD*** *300377*	dystrophin	Muntoni et al. (1993) Milasin et al. (1996)	allelic to DMD (group 1)
Dilated cardiomyopathy due to laminin-alpha4 defect	10.40	AD		6q21	***LAMA4*** *600133*	laminin-alpha4	Knöll et al. (2007)	
Dilated cardiomyopathy due to integrin-linked kinase defect	10.41	AD		11p15.4	***ILK*** *602336*	integrin-linked kinase	Knöll et al. (2007)	
(c) Restrictive								
Restrictive cardiomyopathy, 1	10.42	AD	RCM1 115210	19q34	***TTNI3*** *191044*	Cardiac troponin I	Mogensen et al. (2003)	allelic to CMH7 (group 10)

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Restrictive cardiomyopathy, 2	10.43		RCM2 609578	10	?	?	Zhang et al. (2005)	

Arrhythmogenic and Long QT syndromes (42 items): see online version of the gene table of NMD at http://www.musclegenetable.org

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)
	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 with rapsyn deficiency	11.11	AR	608931	11p11	***RAPSN*** *601592*	rapsyn	Ohno et al. (2002) Ohno et al. (2003) Dunne et al. (2003)	
Congenital myasthenic syndrome with choline acetyltransferase deficiency	11.12	AR	CMS-EA 254210	10q11.2	***CHAT*** *118490*	choline acetyltransferase	Ohno et al. (2001) Maselli et al. (2003)	
Congenital myasthenic syndrome with end-plate acetylcholinesterase deficiency	11.13	AR	EAD 603034	3p24.2	***COLQ*** *603033*	collagen-like tail subunit (single strand of homotrimer) of asymmetric	Donger et al. (1998) Ohno et al. (1998, 1999, 2000)	
Congenital myasthenic syndrome with MuSK deficiency	11.14	AR	CMS1B 608931	9q31-q32	***MUSK*** *601296*	MuSK (Muscle-specific receptor tyrosine kinase)	Chevessier et al. (2004)	
Congenital myasthenic syndrome, type Ia1	11.15	AR	FIM1 605809	17p13	?	?	Christodoulou et al. (1997)	
Familial limb-girdle myasthenia	11.16	AR	LGM 254300	4p16.2	***DOK7*** *610285*	docking protein 7	Beeson et al. (2006); Selcen et al. (2008)	
Sodium-channel myasthenia	11.17	AR	608931	17q23	***SCN4A*** *603967*	sodium channel, voltage-gated, type IV, alpha	Tsujino et al. (2003) [Wendy: please add this ref to list]	allelic to HOKPP2 (group 7), HYPP (group 7), PMC (group 7), K-aggravated myotonia (group 7)
Escobar syndrome (multiple pterygium syndrome)	11.18	AR	265000	2q22-q44	***CHRNG*** *100730*	cholinergic receptor, nicotinic, gamma	Hoffman et al. (2006) Morgan et al. (2006)	
Myasthenic syndrome, myopathy and epidermis bullosa	11.19	AR		8q24-qter	***PLEC1*** *601282*	plectin	Banwell et al. (1999)	allelic to MDEBS (group 5)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
GROUP 12. SPINAL MUSCULAR ATROPHIES								
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 (group 12), SMA3 (group 12), 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 (group 12), SMA2 (group 12), 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 (group 12), SMA2 (group 12.2), SMA3 (group 12.3)
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, juvenile (also classified as "Distal hereditary motor neuropathy type II", in group 14)	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-kDa protein-1	Evgrafov et al. (2004)	allelic to CMT2F (group 14)
Distal spinal muscular atrophy, distal with upper limb predominance (type V)	12.12	AD	DSMAV (HMN5) 600794	7p15	***GARS*** *600287*	glycyl tRNA synthetase	Christodoulou et al. (1995) Antonellis et al. (2003)	allelic to (CMT2D (group14)
Distal spinal muscular atrophy type V	12.13	AD	DSMAV (HMN5) 600794	1q13	***BSCL2*** 606158	seipin	Windpassinger et al. (2004)	allelic to SPG17 (group 15)
Spinal muscular atrophy, distal, with vocal cord paralysis (Harper-Young)	12.14	AD	HMN7A 158580	2q14	*?*	?	McEntagart et al. (2001)	
Distal hereditary motor neuronopathy type VIIB	12.15	AD	HMN7B 607641	2p13	***DCTN1*** *601143*	dynactin 1	Puls et al. (2003)	

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DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Spinal muscular atrophy, distal, X-linked	12.16	XR	SMAX3 300489	Xq13-q21	*?*	?	Takata et al. (2004)	
Spinal muscular atrophy congenital non progressive of lower limbs	12.17		SMAL 600175	12q23-q24	?	?	van der Vleuten et al. (1998)	
Scapuloperoneal spinal muscular atrophy	12.18	AD	SPSMA 181405	12q.24	?	?	Isozumi et al. (1996)	
Others								
Familial amyotrophic lateral sclerosis	12.19	AD	ALS1 105400	21q22	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Siddique et al. (1991, 1996) Rosen et al. (1993)	
Familial amyotrophic lateral sclerosis (recessive)	12.20	AR	ALS1 105400	21q22	***SOD1*** *147450*	Cu/Zn superoxide dismutase	Andersen et al. (1995)	
Amyotrophic lateral sclerosis, juvenile	12.21	AR	ALS2 205100	2q33	***ALS2*** *606352*	alsin	Hentati et al. (1994a) Yang et al. (2001) Hadano et al. (2001)	
Familial amyotrophic lateral sclerosis	12.22	AR	ALS3 606640	18q21	*?*	?	Hand et al. (2002)	
Familial amyotrophic lateral sclerosis	12.23	AD	ALS4 602433	9q34	***SETX*** *608465*	senataxin	Chance et al. (1998) Chen et al. (2004) Moreira et al. (2004)	
Familial amyotrophic lateral sclerosis	12.24	AD	ALS5 602099	15q15-q21	*?*	?	Hentati et al. (1998)	
Familial amyotrophic lateral sclerosis	12.25	AD	ALS6 608030	16q12	*?*	?	Sapp et al. (2003) Abalkhail et al. (2003)	
Familial amyotrophic lateral sclerosis	12.26	AD	ALS7 608031	20p13	*?*	?	Sapp et al. (2003)	
Familial amyotrophic lateral sclerosis	12.27	AD	ALS8 608627	20q13	***VAPB*** *605704*	vesicle-associated membrane protein-associated protein B and C	Nishimura et al. (2004a, 2004b)	
Kennedy disease	12.28	XR	*SBMA 313200*	Xq13	***AR*** *313700*	androgen receptor	Fishbeck et al. (1986) La Spada et al. (1991)	
Lethal Congenital Contracture Syndrome 1	12.29	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	12.30	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	12.31	AR	LCCS3 611359	19p13	***PIP5K1C*** *606102*	phosphatidy-linositol-4-phosphate 5-kinase, type I, gamma	Narkis et al. 2007	

GROUP 13. HEREDITARY ATAXIAS

To See the 48 items of this group please consult the online version of the gene table of NMD at http://www.musclegenetable.org

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 to CMT1E (group14), HNPP (group14), DSS (group14)
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 (group14), CMT2J (group14), DSS (group14), CMT4E (group14)
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 (group14), DSS (group14)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
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 (group14), DSS (group14),
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 (group14) CMT1E (group14), HNPP (group14), DSS (group14)
type 1F	14.7	AD	CMT1F 607734	8p21	***NEFL*** *162280*	neurofilament, light polypeptide 68kDa	Jordanova et al. (2003)	allelic to CMT2E (group14)
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)	
Dominant intermediate (CMTDI)								
type A	14.10	AD	CMTDIA 606483	10q24.1-q25.1	*?*	?	Verhoeven et al. (2001)	
type B	14.11	AD	CMTDIB 606482	19p12-13.2	***DNM2*** *602378*	dynamin 2	Zuchner et al. (2005)	allelic to CNM (group 3)
type C	14.12	AD	CMTDIC 608323	1p35	***YARS*** *603623*	tyrosyl-tRNA synthetase	Jordanova et al. (2003, 2006)	
type D	14.13	AD	CMTDID 607791	1q22	***MPZ*** *159440*	myelin protein zero	Mastaglia et al. (1999)	allelic to CMT1B, CMT4E, CMT2I, CMT2J, DSS (this group)
Autosomal recessive (AR-CMT1 or CMT4)								
CMT, type 4A	14.14	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 (group14)
CMT, type 4B1	14.15	AR	CMT4B1 601382	11q22	***MTMR2*** *603557*	myotubularin-related protein-2	Bolino et al. (1996, 2000) Previtali et al. (2003)	
CMT, type 4B2	14.16	AR	CMT4B2 604563	11p15	***SBF2 (MTMR13)*** *607697*	SET binding factor 2	Azzedine et al. (2003) Senderek et al. (2004)	
CMT, type 4C	14.17	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)	14.18	AR	HMNSL 601455	8q24	***NDRG1*** *605262*	N-MYC downstream regulated gene 1	Kalaydjieva et al. (1996, 2000) Hunter et al. (2003)	
CMT, type 4E (congenital hypomyelinating myopathy)	14.19		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)	14.20		CMT4E 605253	1q22	***MPZ*** *159440*	myelin protein zero	Warner et al. (1996)	allelic to CMT1B (group14), CMT2I (group14), CMT2J (group14), DSS (group14)
CMT, type 4F	14.21	AR	CMT4F 145900	19q13	***PRX*** *605725*	periaxin	Delague et al. (2000) Guilbot et al. (2001)	allelic to DSSE (group14)
CMT, type 4G (type Russe)	14.22	AR	CMT4G 605285	10q22	?	?	Rogers et al. (2000) Thomas et al. (2001)	
CMT, type 4H	14.23	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	14.24	AR	CMT4J 611228	6q21	***FIG4 (KIAA0274)*** *609390*		polyphosphoinositide phosphatase activity	Chow et al. (2007)
X-linked CMT1								
CMT1, X-linked 1	14.25	XD	CMTX1 302800	Xq13	***GJB1*** *304040*	gap junction protein, beta 1, 32kDa (connexin 32)	Bergoffen et al. (1993) Bone et al. (1995)	allelic to DSS (group 14)

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DISEASE NAME	Item line in this group	Inheri-tance	Locus or disease symbol and OMIM number	Chromo-some	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
CMT1, X-linked 2	14.26	XR	CMTX2 302801	Xp22.2	*?*	?	Ionasecu et al. (1992)	
CMT1, X-linked 3	14.27	XR	CMTX3 302802	Xq26	*?*	?	Ionasecu et al. (1992) Huttner et al. (2006)	
CMT1, X-linked 4 (Cowchock syndrome)	14.28	XR	CMTX4 310490	Xq24-q26	*?*	?	Priest et al. (1995)	
CMT1, X-linked 5 (with hearing loss and optic neuropathy	14.29	XR	CMTX5 311070	Xq22-q24	***PRPS1*** *311850*	phosphoribosyl pyrophosphate synthetase 1	Kim at al (2007)	
Dejerine-Sottas syndrome (DSS or CMT3)								
Déjerine-Sottas hypertrophic neuropathy, dominant	14.30	AD	DSSA 145900	17p11.2	***PMP22*** *601097*	peripheral myelin protein 22	Roa et al. (1993b)	allelic to CMT1A (group14) CMT1E (group14), HNPP (group14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.31	AD	DSSB 145900	1q21-q23	***MPZ*** *159440*	myelin protein zero	Hayasaka et al. (1993b)	allelic to CMT1B (group14), CMT2I (group 14), CMT2J (group14), CMT4E (group14)
Déjerine-Sottas hypertrophic neuropathy, dominant	14.32	AD (digenic)	DSSC 145900	10q21.1	***EGR2*** *129010* and ***GJB1*** *304040*	early growth response 2 (Krox-20 homolog) and gap junction protein, beta 1, 32 kDa (connexin 32)	Chung et al. (2005)	allelic to CMTX1 (group14)
Déjerine-Sottas hypertrophic neuropathy, recessive	14.33	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	14.34	AD	CMT2A1 118210	1p36.2	***KIF1B*** *605995*	kinesin family member 1B	Zhao et al. (2001a)	
type 2A2	14.35	AD	CMT2A2 609260	1p36.2	***MFN2*** *608507*	mitofusin 2	Ben Othmane et al. (1993a) Züchner et al. (2004)	
type 2B	14.36	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) Wendy, please add Perikac-Vance et al. in the list]	
type 2C	14.37	AD	CMT2C 606071	12q23-q24	?	?	Klein et al. (2003) McEntagart et al. (2005)	
type 2D	14.38	AD	CMT2D 601472	7p15	***GARS*** *600287*	glycyl tRNA synthetase	Ionasescu et al. (1996) Antonellis et al. (2003)	allelic to DSMAV (group12)
type 2E	14.39	AD	CMT2E 607684	8p21	***NEFL*** *162280*	neurofilament, light polypeptide 68 kDa	Birouk et al. (2003) Claramunt et al. (2005) [Wendy: Please add Birouk ref to the list]	allelic to CMT1F (group14)
type 2F	14.40	AD	CMT2F 606595	7q11-q21	***HSPB1*** *602195*	heat-shock 27-kD protein-1	Ismailov et al. (2001) Evgrafov et al. (2004)	
type 2G	14.41	AD	CMT2G 608591	12q12-q13	*?*	?	Nelis et al. (2004).	
type 2H	14.42	AD	CMT2H 607731	8q21.3	*?*	?	Barhoumi et al. (2001) [Wendy: please add this ref to the list]	maybe allelic to CMT4A (group14)
type 2I (late onset)	14.43	AD	CMT2I 607677	1q22	***MPZ*** *159440*	myelin protein zero	Auer-Grumbach et al. (2003) [Wendy: please add this ref to the list]	allelic to CMT1B (group14), CMT2J (group14), DSS (group14), CMT4E (group14)
type 2J (with hearing loss and pupillary abnormality)	14.44	AD	CMT2J 607736	1q22	***MPZ*** *159440*	myelin protein zero	De Jonghe et al. (1999) Chapon et al. (1999)	allelic to CMT1B (group14), CMT2J (group14), DSS (group14), CMT4E (group14)
type 2K	14.45	AD	CMT2K 607831	8q13-q21	***GDAP1*** *606598*	ganglioside-induced differentiation-associated protein 1	Birouk et al. (2003) Claramunt et al. (2005)	allelic to CMT4A and ARCMT2C (group14)
type 2L	14.46	AD	CMT2L 608673	12q24	***HSPB8*** *608014*	heat shock protein 8	Tang et al. (2004, 2005)	allelic to dHMN (group 14)

DISEASE NAME	Item line in this group	Inheritance	Locus or disease symbol and OMIM number	Chromosome	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Hereditary motor and sensory neuropathy, Okinawa type	14.47	AD	HMSN P 604484	3q13	*?*	?	Takeshima et al. (1997, 1999) add ref 1997 already in list	
CMT2 Autosomal recessive								
Autosomal recessive CMT2A	14.48	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 (group 1), EDMD3 (group1; LGMD1B (group 1) [+FPLD2/151660, HGPS/176670, restrictive dermopathy/275210, not in this table]
Autosomal recessive CMT2B	14.49	AR	CMT2B2 605589	19q13	*?*	*?*	Leal et al. (2001)	
Autosomal recessive CMT2C	14.50	AR	607831	8q13-q21	***GDAP1*** *606598*	ganglioside-induced differentiation-associated protein 1	Nelis et al. (2002) Birouk et al. (2003)	allelic to CMT4A, and CMT2K (group14)
(C) CMT spinal (or HMN) see under SPINAL MUSCULAR ATROPHIES (group 12)								
(D) Other HSMN syndromes								
Hereditary sensory and autonomic neuropathy type I	14.51	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)	
Distal hereditary motor neuropathy type II	14.52	AD	dH*MN2* *158590*	12q24	***HSPB8*** *608014*	heat shock protein 8	Timmerman et al. (1996) Irobi et al. (2004)	allelic to CMT2L (group 14)
Hereditary motor and sensory neuropathy-Lom (with deafness)	14.53	AR	HMNSL 601455	8q24	***NDRG1*** *605262*	Nmyc downstream regulated gene 1	Kalaydjieva et al. (1996, 2000) Hunter et al. (2003)	
Peripheral neuropathy and agenesis of the corpus callosum (Charlevoix disease)	14.54	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)	14.55	AD	HNA 162100	17q25	***SEPT9*** *604061*	septin 9	Pellegrino et al. (1996) Kuhlenbaumer et al. (2005)	
Giant axonal neuropathy	14.56	AR	GAN 256850	16q24.1	***GAN1*** *605379*	gigaxonin	Ben Hamida et al. (1997) Bomont et al. (2000)	
Congenital cataracts, facial dysmorphism and neuropathy	14.57	AR	**CCFDN** 604168	18p23	***CTDP1*** *604927*	CTD phosphatase subunit 1	Varon et al, (2003)	

GROUP 15. HEREDITARY PARAPLEGIAS

To see the 40 items of this group please consult the online version of the gene table of NMD at http://www.musclegenetable.org

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*** *604 149*	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		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)	

(*continued on next page*)

DISEASE NAME	Item line in this group	Inheri-tance	Locus or disease symbol and OMIM number	Chromo-some	Gene symbol and OMIM number	Protein	Key references	Other allelic disease (s) (group in this table)
Congenital fibrosis of the extraocular muscles	16.6	AD	CFEOM2 602078	11q13	***PHOX2A (ARIX)*** *602753*	paired-like aristaless homeobox protein 2A)	Wang et al. (1998) Nakano et al. (2001) [Wendy: please replace Engle (1997) by Wang et al. 1998]	
Congenital fibrosis of the extraocular muscles	16.7	AD	CFEOM3 600638	16q24	?	?	Doherty et al. (1999)	
Distal arthrogryposis type 1	16.8	AD	DA1 108120	9p13	***TPM2*** 190990	tropomyosin 2, â tropomyosin	Sung et al. (2003a)	allelic to NEM4 (group 3), 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-twich 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	troponin T3, fast skeletal 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 2β tropomyosin	Tajsharghi et al. (2007c) Ochala et al. (2007)	NEM4 (group 3) Cap disease (group 3), DA1 (group 16)
Trismus-pseudocamp-todactyly	16.14	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	16.15	AD	PEOA1 157640	15q25	***POLG*** 174763	polymerase, DNA, gamma	Van Goethem et al. (2001)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2;	16.16	AD	PEOA2 609283	4q35	***ANT1*** *103220*	mitochondrial carrier, adenine nucleotide translocator. ANT1	Kaukonen et al. (2000)	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 3	16.17	AD	PEOA3 609286	10q11	***C10orf2 =PE01*** *606075*	T7-like mitochondrial DNA helicase	Suomalinen et al. (1997) Spelbrink et al. (2001)	allelic to IOSCA (group 13)
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4	16.18	AD	PEOA4 610131	10q24	***POLG2*** 604983	polymerase, DNA, gamma-2; POLG accessory subunit; POLGB	Longley et al. (2006)	
Mitochondrial DNA depletion myopathy	16.19	AR	609560	16q22	***TK2*** 188250	thymidine kinase, mitochondrial	Saada et al. (2001)	
Mitochondrial DNA depletion myopathy, encephalo-myopathic form	16.20	AR	609560	13q12-q13	***SUCLA2*** *603921*	succinate-CoA ligase, ADP-forming, beta subunit	Elpeleg et al. (2005)	
Mitochondrial DNA depletion myopathy	16.21	AR	609560	8q23	***RRM2B*** *604712*	ribonucleotide reductase, M2B	Bourdon et al. (2007)	
Progressive external ophthalmoplegia with optic atrophy, optic atrophy 1 with deafness	16.22	AD	125250	3q28-q29	***OPA1*** 605290	OPA1 protein, dynamin related GTPase	Amati-Bonneau et al. (2008) Hudson et al. (2008)	

REFERENCES (added since the last version of the gene table in January 2008. The complete list of key references is on the online version (http://www.musclegenetable.org) where it can be retrieved alphabetically, per item or per group.

Group 1. Muscular dystrophies

Biancheri R, Falace A, Tessa A, Pedemonte M, Scapolan S, Cassandrini D, Aiello C, Rossi A, Broda P, Zara F, Santorelli FM, Minetti C and Bruno C. POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes. Biochem Biophys Res Commun 2007;363:1033–7.

Bolduc V, Thiffault I, Tétreault, M, Dicaire J, Robitaille Y, Jarry J, Rioux MF, Loisel L., Mathieu J, Bouchard JP and Brais B. LGMD2L is caused by mutations in the skeletal muscle highly expressed transmembrane TMEM16E gene. WMS 2008 meeting, abstract (LBN01).

Clement EM, Godfrey C, Tan J, Brockington M, Torelli S, Feng L, Brown SC, Jimenez-Mallebrera C, Sewry CA, Longman C, Mein R, Abbs S, Vajsar J, Schachter H and Muntoni F. Mild POMGnT1 mutations underlie a novel limb-girdle muscular dystrophy variant. Arch Neurol 2008;65:137–41.

Godfrey C, Clement E, Mein R, Brockington M, Smith J, Talim B, Straub V, Robb S, Quinlivan R, Feng L, Jimenez-Mallebrera C, Mercuri E, Manzur AY, Kinali M, Torelli S, Brown SC, Sewry CA, Bushby K, Topaloglu H, North K, Abbs S and Muntoni F. Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain 2007;130:2725–35.

Gueneau L. Bertrand A, Jais JP. Salih MA, Stojkovic T, Wehnert M, Sartoh S, Verhuren A, Beuvin M, Lacene E, Heath S. Zelenika D, Voit T, Eymard B, Ben Yaou Rand Bonne C. A new X-linked form of Emery-Drelfuss Muscular Dystrophy is caused by FHL 1 gene mutations that lead to abnormal muscle differentiation. WMS 2008 abstract (LBNO5).

Group 2. Congenital muscular dystrophies

Van Reeuwjjk J, Grewal PK, Salih MA, Beltran-Valero de Bernabe D, McLaughlan JM, Michielse CB, Herrmann R, Hewitt JE, Steinbrecher A, Seidahmed MZ, Shaheed MM, Abomolha A, Brunner HG, van Bokhoven H and Voit T. Intragenic deletion in the LARGE gene causes Walker-Warburg syndrome. Hum Genet 2007; 121:685-90.

Group 3. Congenital myopathies

Compton A, Albrecht DE, Cooper ST, Mowat D, Jones KJ, Yang N, Seto J, Ranscht B, Bahlo M, Froehner SC and North KN. Mutations in contactin-1, a novel neuromuscular junction protein, cause lethal congenital myopathy associated with sarcolemmal loss of syntrophin and alpha-dystrobrevin. Am J Hum Genet 2008; in press.

Schoser BG, Frosk P, Engel AG, Klutzny U, Lochmuller H and Wrogemann K. Commonality of TRIM32 mutation in causing sarcotubular myopathy and LGMD2H. Ann Neurol 2005;57:591–5.

Tajsharghi H, Leren, TP, Brunvand L, Dahl HM, Tulinius M and Oldfors AT. New congenital myopathy associated with a mutation in the cardiac myosin-binding protein C gene MYBPC3. WMS 2008 abstract (LBN07).

Group 4. Distal myopathies

Fischer D, Hérasse M, Bitoun M, Barragan-Campos HM, Chiras J, Laforet P, Fardeau M, Eymard B, Guicheney P and Romero NB. Characterization of the muscle involvement in dynamin 2-related centronuclear myopathy. Brain 2006;129:1463–69.

Group 5. Other myopathies

Munteanu I, Ramachandran N, Wang P, P A, Rilstone J, Israelian N, T N, Paroutis P, Guo R, ZP R, Nishino I, Chabrol6 B, Pellissier J, Minetti C, Udd B, Fardeau M, Kissel J, Kalimo H, Levy N, Manelson M, Ackerley C and Minassian B. Massive downregulation of the V-ATPase by hypomorphic alleles of the VMA21 gene causes an autophagic myopathy. WMS meeting 2008 abstract (LBN04).

Quinzii CM, Vu TH, Min KC, Tanji K, Barral S, Grewal RP, Kattah A, Camano P, Otaegui D, Kunimatsu T, Blake DM, Wilhelmsen KC, Rowland LP, Hays AP, Bonilla E and Hirano M. X-linked dominant scapuloperoneal myopathy is due to a mutation in the gene encoding four-and-a-half-LIM protein 1. Am J Hum Genet 2008;82:208–13.

Selcen D, Muntoni F, Burton B, Pegoraro E, Sewry C, Bite A and Engel AG. Mutation in BAG3 defines severe novel muscular dystrophy of childhood. Ann Neurol 2008; in press.

Windpassinger C, Schoser B, Straub V, Hochmeister S, Noor A, Lohberger B, Farra N, Petek E, Schwarzbraun T, Ofner L, Loscher WN, Wagner K, Lochmuller H, Vincent JB and Quasthoff S. An X-linked myopathy with postural muscle atrophy and generalized hypertrophy, termed XMPMA, is caused by mutations in FHL1. Am J Hum Genet 2008;82:88–99.

7. Ion channel muscle diseases

Miller TM, Dias da Silva MR, Miller HA, Kwiecinski H, Mendell JR, Tawil R, McManis P, Griggs RC, Angelini C, Servidei S, Petajan J, Dalakas MC, Ranum LP, Fu YH and Ptáèek LJ. Correlating phenotype and genotype in the periodic paralyses. Neurology 2004;63:1647–55.

Ptáček LJ, Trimmer JS, Agnew WS, Roberts JW, Petajan JH and Leppert M. Paramyotonia congenita and hyperkalemic periodic paralysis map to the same sodium-channel gene locus. Am J Hum Genet 1991;49:851–4.

Ptáček LJ, George AL, Jr., Barchi RL, Griggs RC, Riggs JE, Robertson M and Leppert MF. Mutations in an S4 segment of the adult skeletal muscle sodium channel cause paramyotonia congenita. Neuron 1992a;8:891–7.

Ptáček LJ, Tawil R, Griggs RC, Storvick D and Leppert M. Linkage of atypical myotonia congenita to a sodium channel locus. Neurology 1992b;42:431–3.

Group 10. Hereditary cardiomyopathies

See website http : //www.musclegenetable.org

Group 11. Congenital myasthenic syndromes

Banwell BL, Russel J, Fukudome T, Shen XM, Stilling G and Engel AG. Myopathy, myasthenic syndrome, and epidermolysis bullosa simplex due to plectin deficiency. J Neuropathol Exp Neurol 1999;58:832–46.

Selcen D, Milone M, Shen XM, Harper CM, Stans AA, Wieben ED and Engel AG. Dok-7 myasthenia: phenotypic and molecular genetic studies in 16 patients. Ann Neurol 2008;64:71–87.

Group 12. Spinal muscular atrophies

Makela-Bengs P, Jarvinen N, Vuopala K, Suomalainen A, Ignatius J, Sipila M, Herva R, Palotie A and Peltonen L. Assignment of the disease locus for lethal congenital contracture syndrome to a restricted region of chromosome 9q34, by genome scan using five affected individuals. Am J Hum Genet 1998;63:506–16.

Narkis G, Ofir R, Manor E, Landau D, Elbedour K and Birk OS. Lethal congenital contractural syndrome type 2 (LCCS2) is caused by a mutation in ERBB3 (Her3), a modulator of the phosphatidylinositol-3-kinase/Akt pathway. Am J Hum Genet 2007;81:589–95.

Narkis G, Ofir R, Landau D, Manor E, Volokita M, Hershkowitz R, Elbedour K and Birk OS. Lethal contractural syndrome type 3 (LCCS3) is caused by a mutation in PIP5K1C, which encodes PIPKI gamma of the phosphatidylinositol pathway. Am J Hum Genet 2007;81:530–9.

Nousiainen HO, Kestila M, Pakkasjarvi N, Honkala H, Kuure S, Tallila J, Vuopala K, Ignatius J, Herva R and Peltonen L. Mutations in mRNA export mediator GLE1 result in a fetal motoneuron disease. Nat Genet 2008;40:155–7.

Group 13. Hereditary ataxias

See website http : //www.musclegenetable.org

Group 14. Hereditary motor and sensory neuropathies

Azzedine H, Bolino A, Taieb T, Birouk N, Di Duca M, Bouhouche A, Benamou S, Mrabet A, Hammadouche T, Chkili T, Gouider R, Ravazzolo R, Brice A, Laporte J and LeGuern E. Mutations in MTMR13, a new pseudophosphatase homologue of MTMR2 and Sbf1, in two families with an autosomal recessive demyelinating form of Charcot-Marie-Tooth disease associated with early-onset glaucoma. Am J Hum Genet 2003;72:1141–53.

Delague V, Bareil C, Tuffery S, Bouvagnet P, Chouery E, Koussa S, Maisonobe T, Loiselet J, Megarbane A and Claustres M. Mapping of a new locus for autosomal recessive demyelinating Charcot-Marie-Tooth disease to 19q13.1–13.3 in a large consanguineous Lebanese family: exclusion of MAG as a candidate gene. Am J Hum Genet 2000;67:236–43.

Huttner IG, Kennerson ML, Reddel SW, Radovanovic D and Nicholson GA. Proof of genetic heterogeneity in X-linked Charcot-Marie-Tooth disease. Neurology 2006;67:2016–21.

Ionasescu VV, Trofatter J, Haines JL, Summers AM, Ionasescu R and Searby C. X-linked recessive Charcot-Marie-Tooth neuropathy: clinical and genetic study. Muscle Nerve 1992;15:368–73.

Jordanova A, Thomas FP, Guergueltcheva V, Tournev I, Gondim FA, Ishpekova B, De Vriendt E, Jacobs A, Litvinenko I, Ivanova N, Buzhov B, De Jonghe P, Kremensky I and Timmerman V. Dominant intermediate Charcot-Marie-Tooth type C maps to chromosome 1p34–p35. Am J Hum Genet 2003;73:1423–30.

Jordanova A, Irobi J, Thomas FP, Van Dijck P, Meerschaert K, Dewil M, Dierick I, Jacobs A, De Vriendt E, Guergueltcheva V, Rao CV, Tournev I, Gondim FA, D'Hooghe M, Van Gerwen V, Callaerts P, Van Den Bosch L, Timmermans JP, Robberecht W, Gettemans J, Thevelein JM, De Jonghe P, Kremensky I and Timmerman V. Disrupted function and axonal distribution of mutant tyrosyl-tRNA synthetase in dominant intermediate Charcot-Marie-Tooth neuropathy. Nat Genet 2006;38:197–202.

Kim HJ, Sohn KM, Shy ME, Krajewski KM, Hwang M, Park JH, Jang SY, Won HH, Choi BO, Hong SH, Kim BJ, Suh YL, Ki CS, Lee SY, Kim SH and Kim JW. Mutations in PRPS1, which encodes the phosphoribosyl pyrophosphate synthetase enzyme critical for nucleotide biosynthesis, cause hereditary peripheral neuropathy with hearing loss and optic neuropathy (cmtx5). Am J Hum Genet 2007;81:552–8.

Mastaglia FL, Nowak KJ, Stell R, Phillips BA, Edmondston JE, Dorosz SM, Wilton SD, Hallmayer J, Kakulas BA and Laing NG. Novel mutation in the myelin protein zero gene in a family with intermediate hereditary motor and sensory neuropathy. J Neurol Neurosurg Psychiatry 1999;67:174–9.

Nelis E, Erdem S, Van Den Bergh PY, Belpaire-Dethiou MC, Ceuterick C, Van Gerwen V, Cuesta A, Pedrola L, Palau F, Gabreels-Festen AA, Verellen C, Tan E, Demirci M, Van Broeckhoven C, De Jonghe P, Topaloglu H and Timmerman V. Mutations in GDAP1: autosomal recessive CMT with demyelination and axonopathy. Neurology 2002;59:1865–72.

Priest JM, Fischbeck KH, Nouri N and Keats BJ. A locus for axonal motor-sensory neuropathy with deafness and mental retardation maps to Xq24–q26. Genomics 1995;29:409–12.

Senderek J, Bergmann C, Weber S, Ketelsen UP, Schorle H, Rudnik-Schoneborn S, Buttner R, Buchheim E and Zerres K. Mutation of the SBF2 gene, encoding a novel member of the myotubularin family, in Charcot-Marie-Tooth neuropathy type 4B2/11p15. Hum Mol Genet 2003;12:349–56.

Shrimpton AE, Levinsohn EM, Yozawitz JM, Packard DS, Jr., Cady RB, Middleton FA, Persico AM and Hootnick DR. A HOX gene mutation in a family with isolated congenital vertical talus and Charcot-Marie-Tooth disease. Am J Hum Genet 2004;75:92–6.

Varon R, Gooding R, Steglich C, Marns L, Tang H, Angelicheva D, Yong KK, Ambrugger P, Reinhold A, Morar B, Baas F, Kwa M, Tournev I, Guergueltcheva V, Kremensky I, Lochmuller H, Mullner-Eidenbock A, Merlini L, Neumann L, Burger J, Walter M, Swoboda K, Thomas PK, von Moers A, Risch N and Kalaydjieva L. Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome. Nat Genet 2003;35:185-9.

Verhoeven K, Villanova M, Rossi A, Malandrini A, De Jonghe P and Timmerman V. Localization of the gene for the intermediate form of Charcot-Marie-Tooth to chromosome 10q24.1–q25.1. Am J Hum Genet 2001;69:889–94.

Zuchner S, Noureddine M, Kennerson M, Verhoeven K, Claeys K, De Jonghe P, Merory J, Oliveira SA, Speer MC, Stenger JE, Walizada G, Zhu D, Pericak-Vance MA, Nicholson G, Timmerman V and Vance JM. Mutations in the pleckstrin homology domain of dynamin 2 cause dominant intermediate Charcot-Marie-Tooth disease. Nat Genet 2005;37:289–94.

Group 15. Hereditary paraplegias

See website http : //www.musclegenetable.org

Group 16. Other neuromuscular disorders

Amati-Bonneau P, Valentino ML, Reynier P, Gallardo ME, Bornstein B, Boissiere A, Campos Y, Rivera H, de la Aleja JG, Carroccia R, Iommarini L, Labauge P, Figarella-Branger D, Marcorelles P, Furby A, Beauvais K, Letournel F, Liguori R, La Morgia C, Montagna P, Liguori M, Zanna C, Rugolo M, Cossarizza A, Wissinger B, Verny C, Schwarzenbacher R, Martin MA, Arenas J, Ayuso C, Garesse R, Lenaers G, Bonneau D and Carelli V. OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. Brain 2008;131:338–51.