Acta Myologica • 2010; XXIX: p. 1-20

INVITED REVIEW

Mendelian bases of myopathies, cardiomyopathies, and neuromyopathies

G. Piluso[1], S. Aurino[2], M. Cacciottolo[2], F. Del Vecchio Blanco[1], A. Lancioni[2], I.L. Rotundo[2], A. Torella[1], V. Nigro[1 2 3]

[1]Dipartimento di Patologia Generale, Seconda Università di Napoli, Naples, Italy; [2] Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy; [3] Centro Interdipartimentale Ricerca e Management (CIRM), Seconda Università di Napoli, Naples, Italy

A second genetic revolution is approaching thanks to next-generation DNA sequencing technologies. In the next few years, the 1,000$-genome sequencing promises to reveal every individual variation of DNA. There is, however, a major problem: the identification of thousands of nucleotide changes per individual with uncertain pathological meaning. This is also an ethical issue. In the middle, there is today the possibility to address the sequencing analysis of genetically heterogeneous disorders to selected groups of genes with defined mutation types. This will be cost-effective and safer.

We assembled an easy-to manage overview of most Mendelian genes involved in myopathies, cardiomyopathies, and neuromyopathies. This was entirely put together using a number of open access web resources that are listed below. During this effort we realized that there are unexpected countless sources of data, but the confusion is huge. In some cases, we got lost in the validation of disease genes and in the difficulty to discriminate between polymorphisms and disease-causing alleles. In the table are the annotated genes, their associated disorders, genomic, mRNA and coding sizes. We also counted the number of pathological alleles so far reported and the percentage of single nucleotide mutations.

Legenda

Gene; symbol (OMIM) indicates the official name of the gene, the symbol and the number as in the Online Mendelian Inheritance in Man database (3).

Chrom indicates the chromosomal position assigned by the GRCh37/hg19 release at http://genome.ucsc.edu/cgi-bin/hgGateway.

Gene size indicates the total size in base pairs of the most relevant transcript of the gene (for example, for the dystrophin gene we considered the skeletal muscle promotor and the 427kDa product).

RefSeq (mRNA) is the official number of the most relevant gene product.

Exon no. is the number of exons.

mRNA (ORF) is the length of the mRNA and within brackets the length of the open reading frame also counting the first stop codon.

Disease symbol (OMIM) indicates the official name of the associated disorders, the symbol and the number as in the Online Mendelian Inheritance in Man (3).

Inh indicates the disease inheritance AR = autosomal recessive; AD = autosomal dominat, XR = X-linked recessive, ecc.

Variants (unique) is the number of total variants (unique) reported in the Leiden database (L) Human Genome Mutation Database (H) or other specifica databases (O).

subst% indicates the percentage of substitutions of mutated alleles found in patients: this may be important to address the strategy for mutation scanning.

Databases

1. Alzheimer Disease & Frontotemporal Dementia Mutation Database (http://www.molgen.ua.ac.be/ADMutations/)
2. ARVD/C Genetic Variants Database (http://www.arvcdatabase.info/)
3. The Human Genome Mutation Database (www.hgmd.org)
4. Cardiogenomics: Sarcomere Gene Mutation Screening (http://cardiogenomics.med.harvard.edu/project-detail?project_id=230)
5. FHC Mutation Database (http://www.angis.org.au/Databases/Heart/heartbreak.html)
6. The Gene Connection for the Heart (http://www.fsm.it/cardmoc/)
7. Database on Transthyretin Mutations (http://www.ibmc.up.pt/mjsaraiva/ttrmut.html)
8. The Mutation Database of Inherited Peripheral Neuropathies (IPNMDB) (http://www.molgen.ua.ac.be/CMTMutations/)
9. Leiden Muscular Dystrophy pages (http://www.dmd.nl/)
10. MOONEY Laboratory MutDB (http://mutdb.org/)
11. Online Mendelian Inheritance in Man(OMIM) (http://www.ncbi.nlm.nih.gov/omim/)
12. ALSOD - The Amyotrophic Lateral Sclerosis Online genetic Database (http://alsod.iop.kcl.ac.uk/Als/Index.aspx)
13. The Androgen Receptor Gene Mutations Database WWW Server (http://androgendb.mcgill.ca/)
14. Mutation Database of the Protein Kinase C (http://www.retina-international.org/sci-news/prkcgmut.htm)
15. The DMD mutations database UMD-DMD France (http://www.umd.be/DMD/W_DMD/index.html)
16. Zhejiang University Center for Genetic and Genomic Medicine (http://www.china-hvp.org/LOVD/?select_db=CAV3)
17. The Long QT syndrome Database (http://www.ssi.dk/graphics/html/lqtsdb/lqtsdb.htm)

Address for correspondence: Prof. Vincenzo Nigro, Dipartimento di Patologia Generale, Seconda Università di Napoli, via Luigi De Crecchio 7, 80138 Naples, Italy. Tel./Fax +39 081 5665704 - E-mail: nigro@tigem.it

Gene; symbol (OMIM)	Chrom	Gene size (bp)	RefSeq (mRNA)	Exon no.	mRNA (ORF)	Disease symbol (OMIM)	Inh	Variants (unique)	Subst%
Abhydrolase domain containing 5; ABHD5 (604780)	3p21	27,000	NM_016006	7	5,370 (1,050)	Chanarin-Dorfam syndrome; CDS (275630)	AR	H=20	80%
Acetylcholine receptor cholinergic receptor, nicotinic, alpha1 muscle; CHRNA1 (100690)	2q31.1	16,877	NM_001039523	10	2,094 (1,449)	Myasthenic syndrome, fast-channel congenital; FCCMS (608930)	AD	H=18	89%
						Myasthenic syndrome, slow-channel congenital; SCCMS (601462)	AD		
Acetylcholinesterase collagen-like tail subunit; COLQ (603033)	3p24.2	71,618	NM_005677	17	3,007 (1,368)	Congenital myasthenic syndrome with endplate acetylcholinesterase deficiency; EAD (603034)	AR	H=28	75%
Acid alpha-glucosidase preproprotein; GAA (606800)	17q25.3	18,324	NM_000152	20	3,847 (2,859)	Glycogen storage disease II; GSDII (232300)	AR	L=230 H=284	75%
Actin, alpha 1, skeletal muscle; ACTA1 (102610)	1q42.13	2,849	NM_001100	7	1,509 (1,134)	Myopathy, congenital, with fiber-type disproportion; CFTD (255310)	AD	L=325 (199) H=87	85%
						Nemaline myopathy; NEM3 (161800)	AD		
Actin, alpha, cardiac muscle precursor; ACTC1 (102540)	15q14	7,630	NM_005159	7	3,693 (1,134)	Familial hypertrophic cardiomyopathy, 11; CMH11 (612098)	AD	H=9 O=9	100%
Actinin alpha2; ACTN2 (102573)	1q43	77,789	NM_001103	21	4,528 (2,685)	Cardiomyopathy, dilated, 1AA; CMD1AA (612158)	AD	H=5	
Activin A receptor, type II-like kinase 2; ACVR1 (102576)	2q24.1	138,666	NM 001105	9	3,062 (1,530)	Fibrodysplasia ossificans progressiva; FOB (135100)	AD	H=13	92%
Acyl-Coenzyme A dehydrogenase, very long chain; ACADVL (609575)	17p13.1	5,432	NM_000018	20	2,205 (1,968)	Acyl-CoA dehydrogenase, very long-chain, deficiency of; VLCAD (201475)	AR	H=108	74%
Adipose triglyceride lipase (desnutrin); PNPLA2 (609059)	11p15.5	6,315	NM_020376	9	2071 (1,515)	Neutral lipid storage disease with myopathy; NLSDM (610717)	AR	H=8	
Aldehyde dehydrogenase 3 family, member A2; ALDH3A2 (609523)	17p11.2	28,840	NM_001031806	11	3823 (1,458)	Spastic paraplegia, complicated recessive (Sjögren-Larsson syndrome); SLS (270200)	AR	H=77	57%
Alsin; ALS2 (606352)	2q33.1	80,908	NM_020919	34	6935 (4,794)	Amyotrophic lateral sclerosis 2, juvenile; ALS 2 (205100)	AR	H=12	100%
						Primary lateral sclerosis, juvenile; PLSJ (606353)	AR		
						Spastic paralysis, infantile onset ascending; IAHSP (607225)	AR		
Amylo-1,6-glucosidase, 4-alpha-glucanotransferase; AGL (610860)	1p21	73,900	NM_000642	34	7,368 (4,599)	Glycogen storage disease IIIa; GSDIIIa (232400)	AR	H=91	56%
Androgen receptor; AR (313700)	Xq12	180,245	NM_000044	8	4314 (2,763)	Androgen insensitivity; AIS (300068)	XR	H=363	82%
						Breast cancer, male, with Reifenstein syndrome; DHTR (312300)	XR		
						Prostate cancer; AR (176807)	XR		
						Spinal and bulbar muscular atrophy of Kennedy; SBMA (313200)	XR		

Ankyrin 2; ANK2 (106410)	4q25-q26	334,110	NM_001148	46	14,241 (11,874)	Cardiac arrhythmia, ankyrin-B-related; LQT4 (600919)	AD	H=10 O=15	
Anoctamin 5; ANO5 (608662)	11p14.3	90,192	NM_213599	22	6,661 (2,742)	Limb-Girdle, Muscular dystrophy, type 2L; LGMD2L (611307)	AR	L=58 (28) H=2	82%
Aprataxin; APTX (606350)	9p13.3	29,018	NM_175073	9	2,086 (1,029)	Ataxia, Early-Onset, with Oculomotor Apraxia and Hypoalbuminemia; EAOH (208920)	AR	H=15	80%
Ataxin 1; ATXN1 (601556)	6p22.3	462,378	NM_000332	8	10,636 (2,448)	Spinocerebellar Ataxia 1; SCA1 (164400)	AD	CAG repeat expansion in coding seq	
Ataxin 10; ATXN10	22q13.31	173,142	NM_013236	12	3,340 (1,428)	Spinocerebellar ataxia 10; SCA10 (603516)	AD	ATTCT repeat expansion in intron 9	
Ataxin 2; ATXN2 (601517)	12q24.1	147,462	NM_002973	25	4,712 (3,942)	Spinocerebellar Ataxia 2; SCA2 (183090)	AD	CAG repeat expansion in coding seq	
Ataxin 3; ATXN3 (607047)	14q32.12	48,069	NM_004993	11	6923 (1,086)	Spinocerebellar Ataxia 3; SCA3 (109150)	AD	CAG repeat expansion in coding seq	
Ataxin 7; ATXN7 (607640)	3p14.1	138,902	NM_000333	13	7242 (2,679)	Spinocerebellar Ataxia 7; SCA7 (164500)	AD	CAG repeat expansion in coding seq	
Ataxin 8 opposite strand; ATXN8OS (613289)	13q21.33	32,541	NR_002717	5	1,472 (0)	Spinocerebellar Ataxia 8; SCA8 (608768)	AD	'CTG*CAG' repeat expansion in opposite genes	
Atlastin GTPase 1; ATL1 (606439)	14q22.1	73,040	NM_015915	14	2,652 (1,677)	Spastic paraplegia 3 (Strumpell disease); SPG3A (182600)	AD	H=27	
ATPase, Ca++ transporting, fast twitch 1; ATP2A1 (108730)	16q12	26,000	NM_004320	23	3,570 (2,985)	Brody myopathy; 601003	AR	H=8	
ATP-binding cassette, sub-family C (member 9); ABCC9 (601439)	12p12.1	139,303	NM_020297	38	8,312 (4,650)	Cardiomyophaty, dilated, 1o; CMD1O (608569)	AD	H=5	
BCL2-associated athanogene 3; BAG3 (603883)	10q26.11	26,446	NM_004281	4	2,608 (1,728)	Myofibrillar myopathy with BAG3 defect; -612954	AD	L=14 (9) H=1	79%
Berardinelli-Seip congenital lipodystrophy 2 (seipin); BSCL2 (606158)	11q12.3	19,300	NM_001130702	11	2,014 (1,389)	Neuronopathy, distal hereditary motor, type V; HMN5 (600794)	AD	H=21	48%
						Spastic paraplegia 17; SPG17 (270685)	AD		
Bridging integrator 1; BIN1 (601248)	2q14.3	59,258	NM_139343	19	2637 (1,782)	Centronuclear myopathy, recessive; 255200	AR		
Calcium channel, voltage-dependent, beta 2 subunit; CACNB2 (600003)	10p12.33-p12.31	401.083	NM_201596	14	4,078 (1,983)	Brugada syndrome 4; BRUGADA SYNDROME 4 (611876)	AD	H=2	
Calcium channel, voltage-dependent, L type, alpha 1C subunit; CACNA1C (114205)	12p13.33	644,700	NM_000719	47	13,480 (6,417)	Timothy syndrome/ Brugada syndrome 3; TS (601005)/BRUGADA SYNDROME (611875)	AD	H=4	
Calcium channel, voltage-dependent, L type, alpha 1S subunit; CACNA1S (114208)	1q31-q32	73,054	NM_000069	44	6,168 (5,622)	Hypokalaemic periodic paralysis; HOKPP1 (170400)	AD	L=86 (14)	100%
						Malignant hyperthermia susceptibility 5; MHS5 (601887)	AD		
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit; CACNA1A (601011)	19p13	300,020	NM_001127221	47	8,626 (6,786)	Acetazolamide-responsive hereditary paroxymal cerebellar ataxia; APCA/ EA2 (108500)	AD	L=101 (88)	98
Calpain 3; CAPN3 (114240)	15q15.1	52,818	NM_000070	24	3,316 (2,466)	Limb-Girdle, Muscular dystrophy, type 2A; LGMD2A (253600)	AR	L=1,848 (452) H=258	60%

Calsequestrin 2 (cardiac muscle); CASQ2 (114251)	1p13.1	68,799	NM_001232	11	2,527 (1,200)	Ventricular tachycardia, catecholaminergic polymorphic, 2; CPVT2 (611938)	AR	H=11	
Carboxy-terminal domain, RNA polymerase II, polypeptide A phosphatase, subunit 1; CTDP1 (604927)	18q23	74,707	NM_004715	13	3,775 (2,886)	Congenital cataracts, facial dysmorphism and neuropathy; CCFDN (604168)	AR	H=1	
Carnitine palmitoyltransferase II; CPT2 (600650)	1p32.3	17,767	NM_000098	5	3,092 (1,977)	Carmitine palmitoyltransferase deficiency; CPT2 (255110)	AR	H=72	75%
Carnitine-acylcarnitine translocase; SLC25A20 (212140)	3p21.31	42,046	NM_000387	9	1,219 (906)	Carnitine acylcarnitine translocase deficiency; CACT Deficiency (212138)	AR	H=27	56%
Caveolin 3; CAV3 (601253)	3p25.3	12,955	NM_001234	2	1,420 (456)	Distal myopathy with caveolin defect;	AD	L=319 (64) H=27 O=17 (14)	92%
						Familial hypertrophic cardiomyopathy, 13; CMH (192600)	AD		
						Hyperckemia, idiopathic; CPK Elevated (123320)	AD		
						Limb-Girdle, Muscular dystrophy, type 1C; LGMD1C (607801)	AD		
						Long QT syndrome 9; LQT9 (611818)	AD		
						Rippling muscle disease, recessive; RMD (606072)	AR		
						Rippling muscle disease; RMD2 (606072)	AD		
Chaperone-activity of bc1 complex homolog; CABC1 (606980)	1q42.13	47,308	NM_020247	15	2,924 (1,944)	Spinocerebellar Ataxia 9; SCAR9 (612016) Coenzyme q10 deficiency (607426)	AR		
Chloride channel 1, skeletal muscle; CLCN1 (118425)	7q34	35,879	NM_000083	23	3,093 (2,967)	Myotonia congenita, autosomal recessive, Becker disease; MCR (255700)	AR	H=134	89%
						Myotonia dominant (Thomsen disease); THD (160800)	AD		
Choline acetyltransferase isoform; CHAT (118490)	10q11.23	51,997	NM_020986	15	2,124 (1,893)	Myasthenic syndrome, congenital, associated with episodic apnea; CMS-EA (254210)	AR	H=15	
Cholinergic receptor, nicotinic, beta 1 muscle; CHRNB1 (100710)	17p13.1	12,526	NM_000747	11	2,436 (1,506)	Myasthenic syndrome, congenital, Ie, included -; CMS1E (608931)	AR	H=5	
						Myasthenic syndrome, slow-channel congenital; SCCMS (601462)	AR		
Cholinergic receptor, nicotinic, gamma; CHRNG (100730)	2q37.1	6,602	NM_005199	12	2,187 (1,554)	Escobar syndrome; CHRNG (265000)	AR	L=34 (25) H=14	50%

Cholinergic receptor, nicotinic, delta; CHRND (100720)	2q37.1	9,283	NM_000751	12	1,740 (1,554)	Myasthenic syndrome, fast-channel congenital; FCCMS (608930)	AD	H=4	
						Myasthenic syndrome, slow-channel congenital; SCCMS (601462)	AD		
Cholinergic receptor, nicotinic, epsilon; CHRNE (100725)	17p13.2	5,306	NM_000080	12	2,460 (1,482)	Myasthenic syndrome, fast-channel congenital; FCCMS (608930)	AD/ AR	H=89	56%
						Myasthenic syndrome, slow-channel congenital; SCCMS (601462)	AD/ AR		
Chromosome 10 open reading frames; C10orf2 (606075)	10q24.31	6,866	NM_021830	5	3,621 (2,055)	Progressive External Ophthalmoplegia with Mitochondrial Deletion 3; PEOA3 (609286)	AD	H=19	
						Spinocerebellar Ataxia infantile-onset; IOSCA (271245)	AR		
Cofilin 2 (muscle); CFL2 (601443)	14q13.1	3,436	NM_021914	4	3,125 (501)	Nemaline myopathy; NEM7 (610687)	AR	L=17 (11)	13
Collagen, type VI, alpha 1; COL6A1 (120220)	21q22.3	23,301	NM_001848	35	4,225 (3,087)	Bethlem myopathy; 158810	AD	L=92 (41) H=28	89%
						Ullrich syndrome; UCMD (254090)	AR		
Collagen, type VI, alpha 2; COL6A2 (120240)	21q22.3	34,731	NM_001849	28	3,439 (3,060)	Bethlem myopathy; 158810	AD	L=145 (55) H=31	91%
						Ullrich syndrome; UCMD (254090)	AR		
Collagen, type VI, alpha 3; COL6A3 (120250)	2q37.3	90,196	NM_004369	44	10,581 (9,534)	Bethlem myopathy (recessive); 158810	AR	L=218 (47) H=20	93%
						Bethlem myopathy; 158810	AD		
						Ullrich syndrome; UCMD (254090)	AR		
Connexin 40; GJA5 (121013)	1q21.2	17,153	NM_005266	2	3,176 (1,077)	Atrial fibrillation, familial, 1; ATFB1 (608583)	AD	H=2	
Contactin 1; CNTN1 (600016)	12q12	377,737	NM_001843	24	3,427 (3,057)	Congenital lethal myopathy; 612540	AR		214
Crystallin, alpha B; CRYAB (123590)	11q23.1	3,124	NM_001885	3	691 (528)	Cataract, posterior polar 2; CRYA2(123590)	AD	L=47 (19) H=13	76%
						Myofibrillar myopathy, alpha-B crystallin related; MFM (608810)	AD		
Cysteine and glycine-rich protein 3 (cardiac LIM protein); CSRP3 (600824)	11p15.1	20,012	NM_003476	6	1,341 (585)	Cardiomyopathy, dilated, 1m; CMD1M (607482)	AD	H=13	
Desmin; DES (125660)	2q35	8,361	NM_001927	9	2,246 (1,413)	Cardiomyopathy, dilated, 1l -; CMD1I (604765)	AD	L=202 (57) H=52	89%
						Myopathy Myofibrillar Desmin related; DRM (601419)	AD		
Desmocollin 2; DSC2 (125645)	18q12.1	36,445	NM_024422	16	5,197 (2,706)	Arrhythmogenic right ventricular dysplasia, 11; ARVD11 (610476)	AD	L=64 (60) H=3 O=69	61%
Desmoglein 2; DSG2 (125671)	18q12.1	50,787	NM_001943	16	5,652 (3,357)	Arrhythmogenic right ventricular dysplasia, 10; ARVD10 (610193)	AD	L=113 (92) H=23 O=107	78%

Desmoplakin; DSP (125647)	6p24.3	45,077	NM_004415	24	9,730 (8,616)	Arrhythmogenic right ventricular dysplasia, familial, 8; ARVD8 (607450)	AD	H=35 O=5	83%
Docking protein 7; DOK7 (610285)	4p16.3	31,176	NM_173660	7	2,565 (1,515)	Familial limb-girdle myasthenia; LGM (254300)	AR	L=129 (34) H=19	15%
Dynactin 1; DCTN1(601143)	2p13.1	19,194	NM_004082	32	4,492 (3,837)	Neuronopathy, distal hereditary motor, type VIIB; HMN7B (607641)	AD	H=5 O=1	100%
Dynamin 2; DNM2 (602378)	19p13.2	113,825	NM_001005360	21	3633 (2,613)	Centronuclear myopathy, dominant; CNM (160150)	AD	H=7 O=6	71%
						Charcot-Marie-Tooth neuropathy, dominant intermediate B; CMTDIB (606482)	AD		
						Congenital muscular dystrophy with dynamin 2 defect;	AD		
						Dynamin2 related distal myopathy; CNM (160150)	AD		
Dysferlin; DYSF (603009)	2p13.2	220,061	NM_001130987	56	6,771 (6,360)	Distal recessive myopathy (Miyoshi myopathy); MMD1 (254130)	AR	L=1122 (415) H=151	68%
						Muscular dystrophy, limb-girdle, type 2B; LGMD2B (253601)	AR		
						Myopathy, distal, with anterior tibial onset; DMAT (606768)	AR		
Dystrobrevin, alpha; DTNA (601239)	18q12.1	398,555	NM_032975	22	6,522 (2,061)	Left ventricular noncompaction with congenital heart defects; LVNC with CHD (606617)	AD	H=1	
						Left ventricular noncompaction, familial isolated; LVNC (604169)	AD		
Dystrophin; DMD (300377)	Xp21.2-p21.1	2,092,329	NM_004006	79	13,993 (11,058)	Dilated cardiomyopathy due to dystrophin defect; XLCM; CMD3B (302045)	XR	L=21,361 (3,793) H=1,070 O=2,898	71% of small mut. 14.9%
						Duchenne/Becker Muscular Dystrophies; BMD (300376); DMD (310200)	XR		
Early growth response 2 protein; EGR2 (129010)	10q21.3	4,367	NM_000399	2	2,974 (1,431)	Charcot-Marie-Tooth disease, type 1D; CMT1D (607668)	AD	H=10 O=12	95%
						Charcot-Marie-Tooth neuropathy Type 4E; CMT4E (605253)	AD		
						Dejerine-Sottas neuropathy; DSN (145900)	AD		
Electron-transfer-flavoprotein, alpha polypeptide; ETFA (608053)	15q24.2-q24.3	95,182	NM_000126	12	1,352 (1,002)	Multiple acyl-CoA dehydrogenase deficiency; MADD (231680)	AR	H=25	64%
Electron-transfer-flavoprotein, beta polypeptide; ETFB (130410)	19q13.41	21.263	NM_001985	6	915 (768)	Multiple acyl-CoA dehydrogenase deficiency; GAIIB (231680)	AR	H=12	75%

Electron-transferring-flavoprotein dehydrogenase; ETFDH (231675)	4q32.1	36,563	NM_004453	13	2,346 (1,854)	Glutaric aciduria type IIC; GAIIC (231680)	AR	H=70	89%
Emerin; EMD (300384)	Xq28	2,285	NM_000117	6	1,337 (765)	Emery-Dreifuss muscular dystrophy, X-linked , type 1; EDMD1 (310300)	XR	L=324 (106) H=81	41%
Enolase 3, beta muscle specific; ENO3 (131370)	17p13.2	6,037	NM_001976	12	1,521 (1,305)	Glycogen storage disease XIII; GSD13 (612932)	AD	H=2	
Epsilon-sarcoglycan; SGCE (604149)	7q21.3	70,986	NM_001099401	12	1,773 (1,389)	Myoclonus-dystonia syndrome; DYT11 (159900)	AD	L=80 (53) H=39	67%
Eyes absent 4; EYA4 (603550)	6q23.2	290,763	NM_004100	20	5,692 (1,920)	Cardiomyophaty, dilated, 1J; CMD1J (605362)	AD	H=8	
						Deafness, autosomal dominant 10; DFNA10 (601316)	AD		
Fibroblast growth factor 14; FGF14 (601515)	13q33.1	680,920	NM_175929	5	2,829 (759)	Spinocerebellar Ataxia 27; SCA27 (609307)	AD	H=2	
Filamin A, alpha (actin binding protein 280); FLNA (300017)	Xq28	26,106	NM_001456	47	8,486 (7,920)	Cardiovalvular dysplasia, X-linked (Myxomatous valvular dystrophy); CVD1 (314400) Frontometaphyseal dysplasia; FMD (305620) FG syndrome 2; FGS2 (300321) Heterotopia, periventricular, X-linked dominant (300049) Otopalatodigital syndrome, type I; OPD1 (311300)	XR	L=413 (83)	80%
Filamin C, gamma (actin-binding protein - 280); FLNC (102565)	7q32.1	28,845	NM_001458	48	9,137 (8,178)	Myopathy miofibrillar filaminC-related; MFM (609524)	AD	L=1 H=2	1
Four and a half LIM domains 1; FHL1 (300163)	Xq26.3	63,960	NM_001159702	8	2,706 (843)	Emery-Dreifuss muscular dystrophy, X-linked, type 2; EDMD6 (300696)	XR	L=64 (22)	82%
						X-linked myopathy with postural muscle atrophy; XMPMA (300696)	XR		
						Scapuloperoneal myopathy; SPM (300695)	XD		
Frataxin; FXN (606829)	9q21.11	43,515	NM_000144	5	7,168 (633)	Friedreich Ataxia 1; FRDA (229300)	AR	H=39 GAA expansion	69%
Fukutin related protein; FKRP (606596)	19q13.32	12,530	NM_024301	4	3,422 (1,488)	Congenital muscular dystrophy and abnormal glycosylation of dystroglycan; MDC1C (606612)	AR	L=536 (142) H=57	94%
						Limb-Girdle, Muscular dystrophy, type 2I; LGMD2I (607155)	AR		
						Muscle-eye-brain disease; MEB (253280)	AR		
						Walker-Warburg syndrome; WWS (236670)	AR		
Fukutin; FKTN (607440)	9q31.2	82,988	NM_001079802	11	7,455 (1,386)	Dilated cardiomyopathy, 1X; CMD1X (611615)	AR	L=141 (45) H=18	53%
						Fukuyama congenital muscular dystrophy; FCMD (253800)	AR		
						Limb-Girdle, Muscular dystrophy, type 2M; LGMD2M (611588)	AR		
						Walker-Warburg syndrome; WWS (236670)	AR		

FYVE RhoGEF and PH domain-containing protein 4; FGD4 (611104)	12p11.21	143,942	NM_139241	17	8,393 (2,301)	Charcot-Marie-Tooth neuropathy Type 4H; CMT4H (609311)	AR	H=3 O=5	
Galactosidase alfa; GLA (300644)	Xq22.1	10,223	NM_000169	7	1,418 (1,290)	Fabry disease; 301500	XR	H=472	73%
Ganglioside-induced differentiation-associated protein 1; GDAP1 (606598)	8q21.11	16,716	NM_018972	6	3,887 (1,077)	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis; CMT4A (=CMT2H) (607706)	AR	H=27 O=28	74%
						Charcot-Marie-Tooth disease, axonal, type 2K; CMT2K (607831)	AR		
						Charcot-Marie-Tooth disease, type 4A; CMT4A (214400)	AR		
Gap junction protein, beta 1; GJB1 (304040)	Xq13.1	2,010	NM_000166	2	1,654 (852)	Charcot-Marie-Tooth neuropathy X-linked 1; CMTX1 (302800)	XD	H=278 O=305	83%
						Déjerine-Sottas hypertrophic neuropathy - digenic with EGR2; o(145900)	XR		
Gigaxonin; GAN (605379)	16q23.2	65,231	NM_022041	11	4,542 (1,794)	Giant axonal neuropathy; GAN (256850)	AR	H=32 O=44	
GLE1: S. Cerevisiae, homolog-like (yeast); (603371)	9q34.11	37,609	NM_001003722	16	3,341 (1,980)	Lethal arthrogryposis with anterior horn cell disease; LAAHD (611890) Lethal congenital contracture syndrome 1; LCCS1 (253310)	AR		
Glucan (1,4-alpha-), branching enzyme; GBE1 (607839)	3p12.2	272,101	NM_000158	16	3,099 (2,109)	Glycogen storage disease type IV; GSDIV (232500)	AR	H=33	
Glycogen phosphorylase; PYGM (608455)	11q13.1	14,326	NM_005609	20	3,615 (2,529)	Glycogen storage disease V; (McArdle disease) GSD5 (232600)	AR	H=131	
Glycogen synthase 3 glycogen synthase 1 (muscle) glycogen synthase 1 (muscle); GYS1 (138570)	19q13.33	25,228	NM_002103	16	3,617 (2,214)	Glycogen storage disease 0, muscle; GSD0b (611556)	AR	H=3	
Glycyl-tRNA synthetase; GARS (600287)	7p14.3	39,466	NM_002047	17	2,747 (2,220)	Charcot-Marie-Tooth disease, axonal, type 2D; CMT2D (601472)	AD	H=4	
						Neuropathy, distal hereditary motor type V; HMNV (600794)	AD		
Heat shock 27kDa protein 1; HSPB1(602195)	7q11.23	1,739	NM_001540	3	896 (618)	Charcot-Marie-Tooth neuropathy, axonal, Type 2F; CMT2F (606595)	AD	H=6	100%
						Neuronopathy, distal hereditary motor, type IIB neuronopathy, distal her; HMN2B (608634)	AD		
Heat shock 22kDa protein 8; HSPB8 (608014)	12q24.23	15,956	NM_014365	3	2,001 (591)	Charcot-Marie-Tooth neuropathy Type 2L; CMT2L (608673)	AD	H=3 O=4	
						Neuropathy, distal hereditary motor, type IIA; HMN2A (158590)	AD		

Heat shock 60k Da protein 1 (chaperonin); HSPD1 (118190)	2q33.1	13,689	NM_199440	12	2,299 (1,722)	Leukodystrophy, hypomyelinating, 4; HLD4 (612233)	AR	H=1 O=2	
						Spastic paraplegia 13; SPG13 (605280)	AD		
Homeobox D10; HOXD10 (142984)	2q31.1	3,178	NM_002148	2	1,803 (1,023)	CMT with Congenital vertical talus; CVT (192950)	AD	H=1	
Hyperpolarization activated cyclic nucleotide-gated potassium channel 4; HCN4 (605206)	15q24.1	49,405	NM_005477	8	7,227 (3,612)	Sick sinus syndrome, dominant; SSS2 (163800) Brugada syndrome 8 (613123)	AD	H=3 O=2	
Immunoglobulin mu binding protein 2; IGHMBP2 (600502)	11q13.3	36,751	NM_002180	15	3,954 (2,982)	Spinal muscular atrophy with respiratory distress; DSMA1 (SMARD1) (604320)	AR	H=45 O=55	78%
Inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein; IKBKAP (603722)	9q31.3	66,808	NM_003640	37	6,113 (3,999)	Familial dysautonomia (Riley-Day syndrome); HSAN3 (223900)	AR	H=3 O=4	
Inositol 1,4,5-triphosphate receptor type 1; ITPR1 (147265)	3p26.1	354,491	NM_001099952	59	10,096 (8,133)	Spinocerebellar Ataxia 15; SCA15 (606658)	AD		
Integrin, alpha 7; ITGA7 (600536)	12q13.2	23,331	NM_002206	25	4,120 (3,414)	Congenital muscular dystrophy with integrin defect; 613204	AR	L=3 (3)	2
Junction plakoglobin; JUP (173325)	17q21.2	32,106	NM_002230	14	3,508 (2,238)	Arrhythmogenic right ventricular dysplasia, familial, 12; ARVD12 (611528)	AD	L=85 (39) H=1 O=15	
						Naxos disease; 601214	AD		
Kinesin family member 1B; KIF1B (605995)	1p36.22	170,896	NM_015074	47	10,579 (5,313)	Charcot-Marie-Tooth disease, type 1A; CMT2A1 (118210)	AD	H=1	
Kinesin family member 21A; KIF21A (608283)	12q12	150,162	NM_017641	37	6,601 (4,686)	Congenital fibrosis of the extraocular muscles; CFEOM1(135700)	AD	H=11	
Kinesin family member 5A; KIF5A (602821)	12q13.3	34,708	NM_004984	29	3,897 (3,099)	Spastic paraplegia 10; SPG10 (604187)	AD	H=4	
L1 cell adhesion molecule; L1CAM (308840)	Xq28	14,426	NM_000425	28	5,026 (3,774)	Spastic paraplegias, X-linked type 1 (MASA syndrome X-linked hydrocephalus); SPG1 (303350), HSAS (307000)	XR	H=179	68%
Lactate dehydrogenase A; LDHA (150000)	11p15.1	13,830	NM_005566	8	2,208 (999)	Glycogen storage disease XI (Lactate dehydrogenase-A deficiency); GSD11 (612933	AR	H=8	

Lamin A/C; LMNA (150330)	1q22	25,418	NM_170707	12	3,225 (1,995)	Charcot-Marie-Tooth neuropathy Autosomal recessive 2B1; CMT2B1 (605588)	AR	L=608 (252) H=142 O=347	80.2%
						Dilated cardiomyopathy, 1A; CMD1A (115200)	AD		
						Emery-Dreifuss muscular dystrophy (autosomal dominant); EDMD2 (181350)	AD		
						Emery-Dreifuss muscular dystrophy (autosomal recessive); EDMD3 (181350)	AR		
						Hutchinson-Gilford progeria syndrome; HGPS (176670)	AD		
						Limb-Girdle, Muscular dystrophy, type 1B; LGMD1B (159001)	AD		
						Lipodystrophy, familial partial, type 2; FPLD2 (151660)	AD		
						Mandibuloacral dysplasia with type A lipodystrophy; MADA (248370)	AR		
Laminin, alpha 2 (Merosin); LAMA2 (156225)	6q22.33	633,423	NM_000426	65	9,690 (9,369)	Muscular dystrophy, congenital merosin-deficient, 1A; MDC1A (607855)	AR	L=550 (266) H=79	67.6%
Acetylglucosaminyl-transferase-like protein; LARGE (603590)	22q12.3	647,354	NM_004737	16	4,200 (2,271)	Muscular dystrophy, congenital, type 1D; MDC1D (608840) Walker-Warburg syndrome (236670)	AR	L=29 (24) H=2	79%
LIM domain binding 3; LDB3 (605906)	10q23.2	67,397	NM_007078	13	5,296 (2,184)	Cardiomyopathy dilated 1C; CMD1C (601493)	AD	L=79 (17) H=5	61%
						myofibrillar myopathy ZASP-related -; MFMZ (609452)	AD		
Lipopolysaccharide-induced TNF alpha factor; LITAF (603795)	16p13.13	39,225	NM_001136473	5	2,565 (459)	Charcot-Marie-Tooth neuropathy Type 1C; CMT1C (601098)	AD	H=9 O=19	
Lysosomal-associated membrane protein 2 precursor; LAMP2 (309060)	Xq24	43,201	NM_002294	9	6,587 (1,233)	Danon disease; Glycogen storage disease IIb; GSD IIb (300257)	XD	L=11 (6) H=35	57%
Member RAS-associated protein RAB7; RAB7 (602298)	3q21.3	88,663	NM_004637	5	2,232 (624)	Charcot-Marie-Tooth neuropathy Type 2B; CMT2B (600882)	AD		
Mitofusin 2; MFN2 (608507)	1p36.22	33,334	NM_014874	19	4,676 (2,274)	Hereditary motor and sensory neuropathy 2A; CMT2A2 (609260)	AD	H=50	96%

Myelin protein zero; MPZ (159440)	1q23.3	5,236	NM_000530	6	1,953 (777)	Charcot-Marie-Tooth disease, dominant intermediate D; CMTDID (607791)	AD	H=128	82%
						Charcot-Marie-Tooth disease, type 1B; CMT1B (118200)	AD		
						Charcot-Marie-Tooth disease, type 2I; CMT2I (607667)	AD		
						Charcot-Marie-Tooth disease, type 2J; CMT2J (607736)	AD		
						Dejerine-Sottas syndrome; DSS (145900)	AD		
						Neuropathy, congenital hypomyelinating; CMT4E (605253)	AD		
Myosin binding protein C, cardiac; MYBPC3 (600958)	11p11.2	21,296	NM_000256	33	4,216 (3,865)	Familial hypertrophic cardiomyopathy, 4; CMH4 (115197)	AD	H=168 C=150	71%
Myosin heavy chain 3; MYH3 (160720)	17p13.1	27,623	NM_002470	40	6,017 (5,823)	Distal arthrogryposis type 2A, Freeman-Sheldon syndrome; DA2A (193700)	AD	H=15	93%
						Distal arthrogryposis type 2B, Sheldon-Hall syndrome; DA2B (601680)	AD		
Myosin heavy chain 6; MYH6 (160710)	14q11.2	26,288	NM_002471	39	5941 (5,820)	Cardiomyopathy, dilated, 1EE; CMD1EE (613252) Cardiomyopathy, familial hypertrophic; CMH14 (613251)	AD	H=7	
Myosin heavy chain 8; MYH8 (160741)	17p13.1	31,626	NM_002472	40	6,038 (5,814)	Trismus-pseudocamptodactyly Syndrome; DA7 (158300) Carney complex variant; (608837)	AD	H=1	
Myosin light chain 2; MYL2 (160781)	12q24.11	9,779	NM_000432	7	827 (501)	Cardiomyopathy, familial hypertrophic 10; MYL2 (608758)	AD		
Myosin light chain 3; MYL3 (160790)	3p21.31	5,617	NM_000258	7	877 (588)	Cardiomyopathy, familial hypertrophic, 8; CMH8 (608751)	AD	H=9 C=5	
Myosin light chain kinase 2; MYLK2 (606566)	20q11.21	15,323	NM_033118	13	2,793 (1,791)	Cardiomyopathy, familial hypertrophic; CMH (192600)	AD		
Myosin heavy chain 7, cardiac muscle, beta; MYH7 (160760)	14q11.2	22,923	NM_000257	40	6,029 (5,808)	Dilated cardiomyopathy, 1S; CMD1S (160760)	AD	H=229	94.7%
						Distal myopathy (Laing); MPD1 (160500)	AD		
						Familial hypertrophic cardiomyopathy, 1; CMH1 (192600)	AD		
						Hyaline body myopathy,dominant (myosin storage myopathy); 608358	AD		
						Myosin storage myopathy and cardiomyopathy, recessive;	AR		

Myostatin, growth/differentiation factor 8; GDF8; MSTN (601788)	2q32.2	7,029	NM_005259	3	2,818 (1,128)	Muscle Hypertrophy (601788)	AD	H=1	
Myotilin; MYOT (604103)	5q31.2	19,995	NM_006790	10	2,271 (1,497)	Distal myopathy with myotilin defect;	AD	L=111 (17) H=7	100%
						Limb-Girdle, Muscular dystrophy, type 1A; LGMD1A (159000)	AD		
						Myopathy myofibrillar (Myotilinopathy); MFM (609200)	AD		
						Spheroid body myopathy; 182920	AD		
Myotonic dystrophy protein kinase; DMPK (605377)	19q13.32	12,840	NM_004409	15	2,863 (1,890)	Myotonic dystrophy (Steinert disease); DM (160900)	AD	CTG repeat expansion in the 3'UTR	
Myotubularin; MTM1 (300415)	Xq28	104,568	NM_000252	15	3,434 (1,812)	Myotubular myopathy 1; MTM1 (310400)	XR	L=382 (236) H=224	68%
Myotubularin-related protein 2; MTMR2 (603557)	11q21	91,326	NM_016156	15	4,593 (1,932)	Charcot-Marie-Tooth disease, type 4B1; CMT4B1(601382)	AR	H=13 O=18	
Nebulin; NEB (161650)	2q23.3	249,147	NM_001164507	182	26,200 (25,578)	Nemaline myopathy 2; NEM2 (256030)	AR	L=172 (94) H=61	57%
Neurofilament, light polypeptide 68kDa; NEFL (162280)	8p21	5,660	NM_006158	4	3,602	Charcot-Marie-Tooth disease, type 1F; CMT1F (607734)	AD	H=30 O=19	
						Charcot-Marie-Tooth disease, type 2E; CMT2E(607684)	AD		
N-myc downstream regulated gene 1; NDRG1(605262)	8q24	60,133	NM_006096	16	3,123 (1,185)	Charcot-Marie-Tooth disease, type 4D; CMT4D (601455)	AR	H=2 O=2	
Non imprinted in Prader-Willi/Angelman syndrome 1; NIPA1 (608145)	15q11.2	43,563	NM_001142275	5	6,386 (765)	Spastic paraplegia 6; SPG6 (600363)	AD	H=4	
O-linked mannose beta 1,2-N-acetylglucosaminyltransferase; POMGNT1 (606822)	1p34.1	9,768	NM_017739	22	2,753 (1,983)	Limb-Girdle, Muscular dystrophy, type 2O; LGMD2O	AR	L=143 (58) H=29	87%
						Muscle-eye-brain disease; MEB (253280)	AR		
						Walker-Warburg syndrome; WWS (236670)	AR		
Optic atrophy 1; OPA1 (605290)	3q28-q29	104,667	NM_015560	31	6,345 (2,883)	Progressive external ophthalmoplegia, optic atrophy 1 with and without deafness; (125250) Optic Atrophy; OPA1 (165500)	AD	H=122	64%
Paired-like homeobox 2a; PHOX2A (602753)	11q13.4	5,099	NM_005169	3	1,716 (855)	Congenital fibrosis of the extraocular muscles; CFEOM2 (602078)	AR	H=6	
Patatin-like phospholipase domain containing 6; PNPLA6 (603197)	19p13.2	27,616	NM_006702	35	4,490 (3,984)	Spastic paraplegia 39; SPG39 (612020)	AR	O=3	

Periaxin; PRX (605725)	19q13	19,600	NM_020956	6	5,533 (444)	Charcot-Marie-Tooth neuropathy Type 4F; CMT4F (145900)	AR	H=10 O=22	72%
Peripheral myelin protein 22; PMP22 (601097)	17p12	35,548	NM_000304	5	1,828 (483)	Charcot-Marie-Tooth neuropathy Type 1A, demyelinating; CMT1A (118220)	AD	H=65 O=69	71%
						Charcot-Marie-Tooth neuropathy Type 1E; CMT1E (118300)	AD		
						Déjerine-Sottas hypertrophic neuropathy; DSN (145900)	AD		
						Hereditary neuropathy with liability to pressure palsies; HNPP (162500)	AD		
Perlecan; HSPG2 (142461	1p36.12	115,013	NM_005529	97	14,287 (13,176)	Dyssegmental dysplasia, Silverman-Handmaker type; DDSH(224410)	AD	H=32	80%
						Schwartz-Jampel Syndrome; SJS1 (255800)	AD		
Phosphatidylinositol-4-phosphate 5-kinase, type I, gamma; PIP5K1C (606102)	19p13.3	70,264	NM_012398	18	5,047 (2,007)	lethal congenital contractural syndrome 3; LCCS3 (611369)	AR		
Phosphofructokinase, muscle; PFKM (610681)	12q13.11	40,532	NM_001166686	25	3,526 (2,343)	Glycogen storage disease VII; GSD VII (232800)	AR	H=17	94%
Phosphoglycerate kinase 1; PGK1 (311800)	Xq21.1	22,657	NM_000291	11	2,421 (1,254)	Phosphoglycerate kinase 1 deficiency; PGK1 (300653)	XR	L=4 H=19	89%
Phosphoglycerate mutase 2 (muscle); PGAM2 (612931)	7p13	2,860	NM_000290	3	2,860 (762)	Glycogen storage disease X; GSD10 (261670)	AR	H=8	
Phospholamban; PLN (172405)	6q22.31	12,145	NM_002667	2	1,713 (159)	Cardiomyophaty, dilated, 1p; CMD1P (609909	AD	H=5	
Phosphoribosyl pyrophosphate synthetase 1; PRPS1 (311850)	Xq22.3	22,602	NM_002764	7	2,155 (957)	Charcot-Marie-Tooth neuropathy X-linked 5 (with hearing loss and optic neuropathy); CMTX5 (311070) Deafness, X-linked 1; DFNX1 (304500)	XR	H=7	
Plakophilin 2; PKP2 (602861)	12p11.21	106,099	NM_001005242	13	4,305 (2,514)	Arrhythmogenic right ventricular dysplasia, familial, 9; ARVD9 (609040)	AD	L=246 (161) H=74	74.4%
Pleckstrin homology domain containing, family G (with RhoGef domain) member 5; PLEKHG5 (611101)	1p36.31	53,918	NM_198681	22	5,221 (3,189)	spinal muscular atrophy, distal, autosomal recessive, 4; DSMA4 (611067)	AR	H=1	
Plectin 1, intermediate filament binding protein 500kDa; PLEC (601282)	8q24.3	61,593	NM_000445	32	14,798 (13,548)	Epidermolysis bullosa simplex associated with late-onset muscular dystrophy; MDEBS (226670)	AR	H=46	52%
						Epidermolysis bullosa simplex, Ogna type; EBS1(131950)	AR		
Poly(A) binding protein, nuclear 1; PABPN1 (602279)	14q11.2	5,971	NM_004643	7	3,075 (921)	Oculopharyngeal muscular dystrophy; OPMD (164300)	AD AR	H=14	7%

Polymerase (DNA directed), gamma 2; POLG2 (604983)	17q23.3	19,281	NM_007215	8	1,592 (1,458)	Progressive external ophthalmoplegia with mitochondrial DNA deletions type 4; PEOA4 (610131)	AD	H=1	
Polymerase (DNA directed), gamma; POLG (174763)	15q26.1	18,490	NM_002693	23	4,447 (3,720)	Mitochondrial Neurogastrointestinal Encephalopathy Syndrome; MNGIE (603041)	AR	H=76	93%
						Progressive External Ophthalmoplegia with mitochondrial DNA deletions; PEOB (258450)	AR		
						Progressive external ophthalmoplegia with mitochondrial DNA deletions type 1; PEOA1 (157640)	AD		
						Sensory Ataxic Neuropathy, Dysarthria and Ophthalmoparesis; SANDO (607459)	AR		
Polyphosphoinositide phosphatase activity; FIG4 (609390)	6q21	134,211	NM_014845	23	3,105 (2,724)	Charcot-Marie-Tooth neuropathy Type 4J; CMT4J (611228)	AR		
Potassium inwardly-rectifying channel J2; KCNJ2 (600681)	17q24.3	10,506	NM_000891	2	5,384 (1,284)	Andersen Cardiodysrhythmic periodic paralysis; LQT7 (170390)	AD	O=39	92%
Potassium voltage-gated channel, Isk-related family, member 1; KCNE1 (176261)	21q22.12	64,625	NM_000219	1	3,320 (390)	Long QT Syndrome 5; LQT5 (176261)	AD	O=9	
Potassium voltage-gated channel, Isk-related family, member 3; KCNE3 (604433)	11q13.4	12,713	NM_005472	3	3,068 (312)	Brugada syndrome 6 (613119)	AD		
Potassium voltage-gated channel, KQT-like subfamily, member 1; KCNQ1 (607542)	11p15.5-p15.4	404,119	NM_000218	16	3,245 (1,650)	Long QT Syndrome 1; LQT1 (192500)	AD	O=90	91%
						Jervell and Lange-Nielsen syndrome 1; JLNS1; (220400)	AD		
Potassium voltage-gated channel, shaker-related subfamily, member 1; KCNA1 (176260)	12p13	8,348	NM_000217	2	7,981 (1,488)	Episodic ataxia, type 1; EA1 (160120)	AD		
Potassium voltage-gated channel, Shaw-related subfamily, member 3; KCNC3 (176264)	19q13.33	13,870	NM_004977	5	3,176 (2,274)	Spinocerebellar Ataxia 13; SCA13 (605259)	AD	H=2	
Presenilin 2; PSEN2 (600759)	1q42.13	25,532	NM_000447	13	2,298 (1,344)	Cardiomyopathy, dilated, 1V, included; CMD1V (600759)	AD	H=20	95%
Presenilin 1; PSEN1 (104311)	14q24.2	87,256	NM_007318	12	6,067 (1,404)	Cardiomyopathy, dilated, 1U, included; CMD1U (104311)	AD	L=1	
Protein kinase C, gamma; PRKCG (176980)	19q13.42	25,435	NM_002739	18	3,128 (2,094)	Spinocerebellar Ataxia 14; SCA14 (605361)	AD	H=23	0%
Protein kinase, AMP-activated, gamma 2 non-catalytic subunit; PRKAG2 (602743)	7q36.1	321,114	NM_016203	16	3,405 (987)	Cardiomyopathy, familial hypertrophic, 6; CMH6 (600858)	AD	H=14	

Protein phosphatase 2 regulatory subunit B, beta isoform; PPP2R2B (604325)	5q32	491,964	NM_181677	11	2,001 (1,299)	Spinocerebellar Ataxia 12; SCA12 (604326)	AD	CAG repeat expansion in coding seq	
Protein-O-mannosyltransferase 1; POMT1 (607423)	9q34.13	20,904	NM_007171	20	3,145 (1,827)	Limb-Girdle, Muscular dystrophy, type 2K; LGMD2K (609308)	AR	L=324 (115) H=28	78%
						Walker-Warburg syndrome; WWS (236670)	AR		
Protein-O-mannosyltransferase 2; POMT2 (607439)	14q24.3	45,927	NM_013382	21	4,872 (2,253)	Limb-Girdle, Muscular dystrophy, type 2N; LGMD2N	AR	L=118 (68) H=5	
						Walker-Warburg syndrome; WWS (236670)	AR		
Proteolipid protein 1; PLP1 (300401)	Xq22.2	16,107	NM_001128834	8	3,133 (729)	Pelizaeus-Merzbacher disease; PMD (312080)	XR	H=128	77%
						Spastic paraplegia 2; SPG2 (312920)	XR		
Rapsyn; RAPSN (601592)	11p11.2	11,416	NM_005055	8	1,664 (1,062)	Myasthenic syndrome, congenital; CMS1D (608931)	AR	H=28	78%
Receptor accessory protein 1; REEP1 (609139)	2p11.2	124,085	NM_001164730	7	3,735 (606)	Spastic paraplegia 31; SPG31 (610250)	AD	H=6	
Ribonucleotide reductase M2 B (TP53 inducible); RRM2B (604712)	8q22.3	34,.618	NM_015713	9	4,932 (1,056)	Mitochondrial DNA depletion myopathy; 612075	AR	H=7	
						Mitochondrial Neurogastrointestinal Encephalopathy Syndrome; MNGIE (603041)	AR		
Ryanodine receptor 1; RYR1 (180901)	19q13.2	153,864	NM_000540	106	15,390 (15,102)	Malignant Hyperthermia Susceptibility to 1; MHS1 (145600)	AD	L=653 (261) H=282	97.7%
						Malignant Hyperthermia Susceptibility to 1; MHS1 (145600)	AR		
						Central Core Disease of Muscle; CCD (117000)	AD		
						Central Core Disease of Muscle; CCD (117000)	AR		
						Multiminicore disease with external ophtalmopegia; 255320	AR		
Ryanodine receptor 2; RYR2 (180902)	1q43	791,587	NM_001035	105	16,365 (14,904)	Arrhythmogenic right ventricular dysplasia, familial, 2; ARVD2 (600996)	AD	H=130	95%
						Ventricular tachycardia, catecholaminergic polymorphic, 1; CPVT1 (604772)	AD		
Sarcoglycan, alpha; SGCA (600119)	17q21.33	9,927	NM_000023	10	1,432 (1,164)	Limb-Girdle, Muscular dystrophy, type 2D; LGMD2D (608099)	AR	L=596 (109) H=50	89.7%
Sarcoglycan, beta; SGCB (600900)	4q12	17,625	NM_000232	6	4,277 (957)	Muscular dystrophy, limb-girdle, type 2E; LGMD2E (604286)	AR	L=345 (79) H=30	68.4%
Sarcoglycan, delta; SGCD (601411)	5q33.3	441,030	NM_000337	9	9,787 (873)	Dilated cardiomyopathy, 1L; CMD1L (606685)	AD	L=76 (37) H=9	77%
						Limb-Girdle, Muscular dystrophy, type 2F; LGMD2F (601287)	AR		

Sarcoglycan, gamma; SGCG (608896)	13q12.12	144,245	NM_000231	8	1,655 (876)	Limb-Girdle, Muscular dystrophy, type 2C; LGMD2C (253700)	AR	L=473 (95) H=25	49.1%
Selenoprotein N, 1; SEPN1 (606210)	1p36.11	18,047	NM_020451	7	4,332 (1,773)	Desmin-related myopathy with Mallory bodies; RSMD1 (602771)	AD	L=198 (61) H=20	76%
						Multiminicore disease, classical form; RSMD1 (602771)	AR		
						Myopathy, congenital, with fiber-type disproportion; CFTD (255310)	AR		
						Rigid spine syndrome; RSMD1 (602771)	AR		
Senataxin; SETX (608465)	9q34.13	93,545	NM_015046	26	11,015 (8,034)	Neuropathy, distal hereditary motor, with pyramidal features; Amyotrophic lateral sclerosis 4, juvenile, ALS4 (602433)	AD	H=27 O=3	77%
						Spinocerebellar ataxia, autosomal recessive 1; SCAR1 (606002)	AR		
Septin 9; SEPT9 (604061)	17q25.2-q25.3	219,185	NM_001113491	12	3,823 (1,761)	Hereditary neuralgic amyotrophy (familial brachial plexus neuropathy); HNA (162100)	AD	H=3 O=3	
Serine palmitoyltransferase long-chain base subunit 1; SPTLC1 (605712)	9q22.31	84,264	NM_006415	15	2,780 (1,422)	Neuropathy, hereditary sensory and autonomic, type 1; HSN1 (162400)	AD	H=4 O=4	
SET binding factor 2; SBF2 (607697)	11p15.3	515,541	NM_030962	40	7,439 (5,550)	Charcot-marie-tooth disease, type 4b2; CMT4B2 (604563)	AR	H=5 O=5	
SH3 domain and tetratricopeptide repeat domain 2; SH3TC2 (608206)	5q32	81,023	NM_024577	17	26,578 (3,867)	Charcot-Marie-Tooth neuropathy Type 4C; CMT4C (601596)	AR	H=19	84%
SIL1 homolog, endoplasmic reticulum chaperone; SIL1 (608005)	5q31.2	251,654	NM_022464	10	1,898 (1,386)	Marinesco-Sjogren Syndrome; MSS (248800)	AR	H=13	61%
Skeletal muscle receptor tyrosine kinase; MUSK (601296)	9q31.3	132.228	NM_005592	14	2,754 (2,610)	Congenital myasthenic syndrome with MuSK deficiency; CMS1B (608931)	AR	H=2	
Slow troponin C; TNNC1 (191040)	3p21.1	2,950	NM_003280	6	695 (486)	Cardiomyopathy, dilated, 1z; CMD1Z (611879)	AD	H=7	
Slow troponin T; TNNT1 (191041)	19q13.4	16.445	NM_003283	14	1,047 (837)	Nemaline Myopathy 5, Amish type; NEM5 (605355)	AR	L=25 (12) H=1	60%
Sodium channel, voltage-gated, type IV, alpha; SCN4A (603967)	17q23.3	34,365	NM_000334	24	7,805 (5,511)	Hyperkalemic periodic paralysis; HYPP (170500) Myotonia, potassium-aggravated (9608390) Hypokalemic periodic paralysis, type 2; HOKPP2 (613345) Myasthenic syndrome, congenital, associated with episodic apnea (254210) Paramyotonia congenita of Von Eulenburg; PMC (168300)	AD	L=613 (101)	99.1%

Solute carrier family 12, (potassium/chloride transporters) member 6; SLC12A6 (604878)	15q14	108,068	NM_001042495	26	7,362 (3,276)	Agenesis of the corpus callosum with peripheral neuropathy; (Charlevoix disease); ACCPN (218000)	AR	H=8 O=5	
Solute carrier family 22 member 5; SLC22A5 (603377)	5q31.1	25,902	NM_003060	10	3,234 (1,674)	Carnitine deficiency, systemic primary; CDSP (212140)	AR	H=68	
Solute carrier family 25 (mitochondrial carrier; adenine nucleotide translocator), member 4; SLC25A4 (103220)	4q35.1	7,121	NM_001151	4	4,419 (897)	Progressive external ophthalmoplegia with mitochondrial DNA deletions type 2; PEOA2 (609283)	AD	H=6 O=7	
Spastic ataxia of Charlevoix-Saguenay (sacsin); SACS (604490)	13q12.12	104,877	NM_014363	10	15,639 (13,740)	Spastic ataxia of Charlevoix-Saguenay; ARSACS (270550)	AR	H=21	48%
Spatacsin; SPG11 (610844)	15q21.1	100,982	NM_025137	40	7,787 (7,332)	Spastic paraplegia 11; SPG11 (604360)	AR	H=10	
Spastic paraplegia 20 (spartin); SPG20 (607111)	13q13.3	44,870	NM_015087	9	5,013 (2,001)	Spastic paraplegia 20 (Troyer); SPG20 (275900)	AR	H=1 O=4	
Spastica paraplegia 21 Acidic cluster protein, 33-kD; SPG21 (608181)	15q22.31	26,888	NM_016630	9	1,797 (927)	Mast syndrome (Spastic paraplegia 21); SPG21 (248900)	AR	H=1	
Spastic paraplegia 7 Paraplegin;SPG7 (602783)	16q24.3	49,369	NM_003119	17	3,081 (2,388)	Spastic paraplegia 7; SPG7 (607259)	AR	H=14	
Spastic paraplegia 4 Spastin; SPG4 (604277)	2p22.3	94,026	NM_014946	17	5,212 (1,851)	Spastic paraplegia 4; SPG4 (182601)	AD	H=237	58%
Synaptic nuclear envelope protein 1; SYNE1 (608441)	6q25.1-q25.2	515,712	NM_182961	146	27,744 (26,394)	Arthrogryposis multiplex congenita with nesprin-1 defect	AR	L=72 (28) H=5	80%
						Autosomal recessive spinocerebellar ataxia, 8; SCAR8 (610743)	AR		
						Emery-Dreifuss with nesprin-1 defect; EDMD4 (612998)	AD		
Synaptic nuclear envelope protein 2; SYNE2 (608442)	14q23.2	373,483	NM_182914	116	21,875 (20,724)	Emery-Dreifuss with nesprin-2 defect; EDMD5 (612999)	AD	L=12 (4) H=1	7
Spectrin, beta, non-erythrocytic 2; SPTBN2 (604985)	11q13.2	36,150	NM_006946	37	7,866 (7,173)	Spinocerebellar Ataxia 5; SCA5 (600224)	AD	H=3	
Strumpellin; KIAA0196 (610657)	8q24.13	67,557	NM_014846	29	4,163 (3,480)	Spastic paraplegia 8; SPG8 (603563)	AD	H=3	
Succinate-CoA ligase, ADP-forming, beta subunit; SUCLA2 (603921)	13q14.2	58,671	NM_003850	11	2,163 (1,392)	Mitochondrial DNA depletion myopathy, encephalomyopathic form with methylmalonic aciduria; (612073)	AR	H=4 O=4	
Superoxide dismutase 1; SOD1 (147450)	21q22.11	9,307	NM_000454	5	963 (465)	Amyotrophic lateral sclerosis 1; ALS1 (105400)	AD	H=138	93%

Tafazzin; TAZ (300394)	Xq28	10,185	NM_000116	11	1,901 (879)	Barth syndrome; BTHS (302060)	XR	H=41	70%
						Cardiomyopathy, X-linked dilated; CMD3A (300069)	XR		
						Endocardial fibroelastosis-2; G4.5 (300394)	XR		
						Noncompaction of left ventricular myocardium, isolated; INVM (300183)	XR		
TATA box binding protein; TBP (600075)	6q27	18,537	NM_003194	8	1,903 (1,020)	Spinocerebellar Ataxia 17; SCA17 (607136)	AD	H=5	
Tau tubulin kinase 2; TTBK2 (611695)	15q15.2	176,466	NM_173500	15	5,621 (3,735)	Spinocerebellar Ataxia 11; SCA11 (604432)	AD		
Thymidine kinase mitochondrial; TK2 (188250)	16q21	42,409	NM_004614	10	5,113 (924)	Mitochondrial DNA depletion myopathy; 609560	AR	H=18	77%
Titin; TTN (188840)	2q31.2	281,431	NM_133378	312	101,516 (100,272)	Autosomal dominant myopathy with proximal muscle weakness and early respiratory muscle involvement (Edstrom myopathy); HMERF (603689)	AD	L=60 (35) H=16	62%
						Congenital myopathy with fatal cardiomyopathy; EOMFC (617105)	AR		
						Dilated cardiomyopathy, 1G; CMD1G (604145)	AD		
						Familial hypertrophic cardiomyopathy, 9; CMH9 (188840)	AD		
						Limb-Girdle, Muscular dystrophy, type 2J; LGMD2J (608807)	AR		
						Tardive tibial muscular dystrophy (Udd myopathy); TMD (600334)	AD		
Titin-cap (Telethonin); TCAP (604488)	17q12	1,208	NM_003673	2	962 (504)	Dilated cardiomyopathy, 1N; CMD1N (607487)	AD	L=53 (28) H=8	75%
						Limb-Girdle, Muscular dystrophy, type 2G; LGMD2G (601954)	AR		
Tocopherol (alpha) transfer protein; TTPA (600415)	8q12.3	26,565	NM_000370	5	2,630 (837)	Ataxia with isolated vitamin E deficiency; TTPA (277460)	AR	H=19	68%
Torsin family 1, member A; TOR1A (605204)	9q34.11	11,220	NM_000113	5	2,105 (999)	Torsion dystonia, early onset; EOTD, DYT1 (128100)	AD	H=4	
Transforming growth factor, beta 3; TGFB3 (190230)	14q24.3	23,651	NM_003239	7	3,183 (1,239)	Arrhythmogenic right ventricular dysplasia; TGFB3 (107970)	AD	L=23 H=2	78%
Transmembrane protein 43; TMEM43 (612048)	3p25.1	18,741	NM_024334	12	3,342 (1,203)	Arrhythmogenic right ventricular dysplasia, familial, 5; ARVD5 (604400)	AD	H=1	
Transthyretin; TTR (176300))	18q12.1	7,255	NM_000371	4	926 (444)	Carpal Tunnel Syndrome; CTS1 (115430)	AD	H=101 O=115	99%
						Dysthyretinemic euthyroidal hyperthyroxinemia; 145680	AD		
						Familiar hereditary amyloidoses; FAP (105210)	AD		

Tripartite motif-containing 32; TRIM32 (602290)	9q33.1	13,999	NM_012210	2	3,719 (1,962)	Limb-Girdle, Muscular dystrophy, type 2H; LGMD2H (254110)	AR	L=27 (11) H=2	73%
						Bardet-Biedl syndrome; BBS (209900)	AR		
Tropomyosin 1 (alpha); TPM1 (191010)	15q22.2	29,274	NM_001018020	9	1,786 (855)	Cardiomyopathy, dilated, 1Y; CMD1Y (611878) Familial hypertrophic cardiomyopathy, 3; CMH3 (115196)	AD	H=13 O=13	100%
Tropomyosin 2 (beta); TPM2 (190990)	9p13.3	7,130	NM_003289	9	1,326 (855)	Cap disease; NEM4 (609285)	AD	L=39 (17) H=3	62%
						Distal arthrogryposis type 1; DA1 (108120)	AD		
						Distal arthrogryposis type 2B, Sheldon-Hall syndrome; DA2B (601680)	AD		
						Nemaline myopathy; NEM4 (609285)	AD		
Tropomyosin 3; TPM3 (191030)	1q21.3	30,320	NM_152263	10	7,096 (747)	Myopathy congenital, with fiber-type disproportion; CFTD (255310)	AR	L=52 (16) H=5	84%
						Nemaline myopathy; NEM1 (609284)	AD		
Troponin I fast-twitch skeletal muscle isoform; TNNI2 (191043)	11p15.5	2,678	NM_003282	7	731 (549)	Distal arthrogryposis type 2B (Sheldon-Hall syndrome); DA2B (601680)	AD	L=26 (16) H=4	53%
Troponin I, cardiac muscle isoform; TNNI3 (191044)	19q13.42	5,964	NM_000363	7	839 (633)	Cardiomyopathy, familial hypertrophic-CMH7; CMH7 (191044)	AD	H=34 O=35	87%
						Cardiomyopathy, familial restrictive - RCM1; RCM1(115210) Cardiomyopathy, dilated, 2A; CMD2A (611880)	AD		
Troponin T3, fast skeletal; TNNT3 (600692)	11p15.5	19,137	NM_006757	16	1,202 (777)	Distal arthrogryposis type 2B (Sheldon-Hall syndrome); DA2B (601680)	AD	L=13 (8) H=1	
Troponin T2, cardiac; TNNT2 (191045)	1q32.1	18,663	NM_000364	14	1,152 (888)	Cardiomyopathy, familial hypertrophic, 2; CMH2 (115195) Cardiomyopathy, dilated, 1D; CMD1D (601494)	AD	H=53 O=31	91%
Tyrosyl-DNA phosphodiesterase 1; TDP1 (607198)	14q32.11	88,861	NM_018319	17	3,745 (1,827)	Spinocerebellar Ataxia with axonal neuropathy; SCAN1 (607250)	AR	H=2	
Tyrosyl-tRNA synthetases; YARS (603623)	1p35.1	42,794	NM_003680	13	3,117 (1,587)	Charcot-Marie-Tooth neuropathy, dominant intermediate C; CMTDIC (608323)	AD	H=3	
UDP-N-acetylglucosamine-2- epimerase/ N-acetylmannosamine kinase; GNE (603824)	9p13.3	62,601	NM_001128227	12	5,283 (2,262)	Inclusion body myopathy 2; IBM2 (600737) Nonaka Myopathy; NM (605820)	AR	H=60	
Vacuolar H+-ATPase homolog; VMA21 (310440)	Xq28	12,179	NM_001017980	3	4,735 (306)	Myopathy with excessive autophagia; MEAX (310440)	XR	L=28 (7) H=6	50%
V-erb-b2 erythroblastic leukemia viral oncogene homolog 3 (avian); ERBB3 (190151)	12q13.2	23,236	NM_001982	28	5,510 (4,029)	lethal congenital contracture syndrome 2; LCCS2 (607598)	AR		

Vesicle-associated membrane protein-associated protein B and C; VAPB (605704)	20q13.32	57,717	NM_004738	6	3,673 (732)	Amyotrophic lateral sclerosis; ALS8 (608627)	AD	H=1	
Vinculin; VCL (193065)	10q22.2	122,041	NM_003373	21	5,280 (3,201)	Cardiomyopathy, familial hypertrophic, 15; CMH15 (613255)	AD	H=1	
Voltage-gated potassium channel, subfamily H, member 2; KCNH2 (152427)	7q36.2	32,965	NM_000238	15	3,896 (3,480)	Long QT Syndrome 2; LQT2 (152427)	AD	O=98	77%
Voltage-gated sodium channel type V alpha; SCN5A (600163)	3p22.2	101,610	NM_198056	28	8,503 (6,051)	Brugada syndrome 1 (601144) Cardiomyopathy, dilated, 1E; CMD1E (601154) Long QT syndrome 3 (603830) Progressive familial heart block, TYPE IA (113900)	AD	O=20	80%
Zinc finger protein 9; CNBP/ ZNF9 (116955)	3q21.3	16,152	NM_001127192	5	3,383 (534)	Myotonic dystrophy; DM2 (602668)	AD	CCTG repeat expansion in intron 1	
Zinc finger, FYVE domain containing 26, spastizin; ZFYVE26 (612012)	14q24.1	70,070	NM_015346	42	9,675 (7,620)	Spastic paraplegia 15 (Kjellin syndrome); SPG15 (270700)	AR	O=4	
Zinc finger, FYVE domain containing 27; protrudin, ZFYVE27 (610243)	10q24.2	23.786	NM_144588	13	3,048 (1,236)	Spastic paraplegia 33; SPG33 (610244)	AD	H=1	