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Disease	Abbreviation	Gene Symbol	Gene Name	Chromosome	MIM#	Key Reference
MUSCULAR DYSTROPHIES						
Duchenne muscular dystrophy	DMD	*DMD*	dystrophin (muscular dystrophy, Duchenne and Becker types)	Xp21.2	310200	Hart (1987) Hum Genet 77, 88
Becker muscular dystrophy	BMD	*DMD*			300376	Monaco (1988) Genomics 2, 90
Limb-girdle muscular dystrophy, type 2A	LGMD2A	*CAPN3*	calpain 3, (p94)	15q15.1	253600	Richard (1995) Cell 81, 27
Limb-girdle muscular dystrophy, type 2B	LGMD2B	*DYSF*	dysferlin, limb girdle muscular dystrophy 2B (autosomal recessive)	2p13.3	253601	Bashir (1998) Nat Genet 20, 37
Limb-girdle muscular dystrophy, type 2C	LGMD2C	*SGCG*	sarcoglycan, gamma	13q12	253700	Noguchi (1995) Science 270, 819
Limb-girdle muscular dystrophy, type 2D	LGMD2D	*SGCA*	sarcoglycan, alpha	17q12-q21.33	608099	Roberds (1994) Cell 78, 625
Limb-girdle muscular dystrophy, type 2E	LGMD2E	*SGCB*	sarcoglycan, beta	4q12	604286	Lim (1995) Nat Genet 11, 257
Limb-girdle muscular dystrophy, type 2L	LGMD2L	*ANO5*	anoctamin 5	11p14.3	609308	Bolduc (2010) Am J Hum Genet 86, 213
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	MDDGC1 (LGMD2K)	*POMT1*	protein-O-mannosyltransferase 1	9q34.1	609308	Godfrey (2007) Brain 130, 2725
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	MDDGC2 (LGMD2N)	*POMT2*	protein-O-mannosyltransferase 2	14q24	613158	Biancheri (2007) Biochem Biophys Res Commun 30, 363
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	MDDGC3 (LGMD2O)	*POMGNT1*	protein O-linked mannose beta1,2-N-acetylglucosaminyltransferase	1p34.1	606822	Clement (2008) Arch Neurol 65, 137
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4	MDDGC4 (LGMD2M)	*FKTN*	fukutin	9q31	611588	Kobayashi (1998) Nature 394, 388
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	MDDGC5 (LGMD2I)	*FKRP*	fukutin related protein	19q13.3	607155	Brockington (2001) Hum Mol Genet 10, 2851
Limb-girdle muscular dystrophy, type 1A	LGMD1A	*MYOT*	myotilin (TTID)	5q31	159000	Hauser (2000) Hum Mol Genet 9, 2141
Limb-girdle muscular dystrophy, type 1B	LGMD1B	*LMNA*	lamin A/C	1q22	159001	Muchir (2000) Hum Mol Genet 9, 1453
Limb-girdle muscular dystrophy, type 1C	LGMD1C	*CAV3*	caveolin 3	3p25	607801	Minetti (1998) Nat Genet 18, 365
Facioscapulohumeral muscular dystrophy (FSHD)	FSHMD1A	*?*	-	4q35	158900	Wijmenga (1990) Lancet. 15, 336
Emery-Dreifuss muscular dystrophy 1, X-linked	EDMD1	*EMD*	Emery-Dreyfuss muscular dystrophy (Emerin)	Xq28	310300	Bione (1994) Nat Genet 8, 323
Emery-Dreifuss muscular dystrophy 2, autosomal dominant	EDMD2	*LMNA*	lamin A/C	1q22	181350	Bonne (1999) Nat Genet 21, 285
Emery-Dreifuss muscular dystrophy 3, autosomal recessive	EDMD3	*LMNA*			604929	
Emery-Dreifuss muscular dystrophy 6, X-linked	EMDD6	*FHL1*	Four and a half LIM domains 1	Xq26	300696	Gueneau (2009) Am J Hum Genet 85, 338
Oculopharyngeal muscular dystrophy	OPMD	*PABPN1*	Poly(A) binding protein, nuclear 1	14q11.2-q13	164300	Brais (1998) Nat Genet 18, 164
CONGENITAL MUSCULAR DYSTROPHIES						
Muscular dystrophy, congenital merosin-deficient, 1C	MDC1A	*LAMA2*	laminin alpha 2 (merosin)	6q22	607855	Helbling-Leclerc (1995) Nat Genet 11, 216
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	MDDGA1	*POMT1*	protein-O-mannosyltransferase 1	9q34.1	236670	Beltran-Valero (2002) Am J Hum Genet 71, 1033
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	MDDGB1	*POMT1*			613155	Bouchet (2007) Hum Mutat 28, 1020
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	MDDGA2	*POMT2*	protein-O-mannosyltransferase 2	14q24	613150	van Reeuwijk (2005) J Med Genet 42, 907
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2	MDDGB2	*POMT2*			613156	
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	MDDGA3	*POMGNT1*	protein O-linked mannose beta1,2-N-acetylglucosaminyltransferase	1p34.1	253280	Yoshida (2001) Developmental Cell 1, 717
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3	MDDGB3	*POMGNT1*			613151	
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	MDDGA4 (FCMD)	*FKTN*	fukutin	9q31	253800	
Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4	MDDGB4	*FKTN*			613152	
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	MDDGA5	*FKRP*			613153	
Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5	MDDGB5 (MDC1C)	*FKRP*	fukutin related protein	19q13.3	606612	Brockington (2001) Am J Hum Genet 69, 1198
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	MDDGA6	*LARGE*			613154	van Reeuwijk (2007) Hum Genet 121, 685
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6	MDDGB6 (MDC1D)	*LARGE*	Like-glycosyltransferase (acetylglucosaminyltransferase-like protein)	22q12.3	608840	Longman (2003) Hum Mol Genet 12, 2853
Muscular dystrophy, rigid spine, 1	RSMD1	*SEPN1*	selenoprotein N, 1	1p36.13	602771	Moghadaszadeh (1998) Am J Hum Genet. 62, 6
Ullrich congenital muscular dystrophy	UCMD	*COL6A1*	collagen VI alpha 1	21q22.3	254090	Pan (2003) Am J Hum Genet 73, 355
Bethlem myopathy		*COL6A1*			158810	Jobsis (1996) Nat Genet 14, 113
Ullrich congenital muscular dystrophy	UCMD	*COL6A2*	Collagen VI alpha 2	21q22.3	254090	Vanegas (2001) Proc Natl Acad Sci U S A 98, 7516
Bethlem myopathy		*COL6A2*			158810	Jobsis (1996) Nat Genet 14, 113
Ullrich congenital muscular dystrophy	UCMD	*COL6A3*	Collagen, type VI, alpha 3	2q37	254090	Demir (2002) Am J Hum Genet 70, 1446
Bethlem myopathy		*COL6A3*			158810	Pan (1998) Hum Mol Genet 7, 807
Muscular dystrophy, congenital, megaconial type	MDCMC	*CHKB*	Choline kinase beta	22q13.33	602541	Mitsuhashi (2011) Am J Hum Genet 88, 845
CONGENITAL MYOPATHIES						
Nemaline myopathy 3	NEM3	*ACTA1*	actin, alpha 1, skeletal muscle	1q42.13-q42.2	161800	Nowak (1999) Nat Genet 23, 208
Myopathy, congenital, with fiber-type disproportion 3	CFTD	*ACTA1*			255310	Laing (2004) Ann Neurol 56, 689
Myopathy, centronuclear, autosomal dominant	CNM1	*DNM2*	dynamin 2	19p13.2	160150	Bitoun (2005) Nat Genet 37, 1207
Myopathy, centronuclear, autosomal recessive	CNM2	*BIN1*	bridging integrator 1	2q14	255200	Nicot (2007) Nat Genet 39, 1134
Myopathy, centronuclear, X-linked	CNMX	*MTM1*	myotubularin 1	Xq27.3-q28	310400	Dahl (1995) Am J Hum Genet 56, 1108
Central core disease of muscle	CCD	*RYR1*	ryanodine receptor 1 (skeletal)	19q13.1	117000	Zhang (1993) Nat Genet 5, 46
Minicore myopathy with external ophthalmoplegia		*RYR1*			255320	
Myopathy, congenital, with fiber-type disproportion	CFTD	*SEPN1*	selenoprotein N, 1	1p36.13	255310	
Nemaline myopathy 1, autosomal dominant	NEM1	*TPM3*	tropomyosin 3 (non-muscle)	1q21.2	609284	Laing (1995) Nat Genet 9, 75
Myopathy, congenital, with fiber-type disproportion	CFTD	*TPM3*			255310	Clarke (2008) Ann Neurol 63, 329
Early-onset myopathy with fatal cardiomyopathy	EOMFC	*TTN*	titin	2q31	611705	Carmignac (2007) Ann Neurol 61, 340
Myofibrillar myopathy, ZASP-related	MFM	*LDB3*	LIM domain binding 3	10q22.3-q23.2	609452	Selcen (2005) Ann Neurol 57, 269
DISTAL MYOPATHIES						
Distal myopathy with decreased caveolin 3		*CAV3*	caveolin 3	3p25	601253	
Miyoshi muscular dystrophy 1	MMD1	*DYSF*	dysferlin, limb girdle muscular dystrophy 2B (autosomal recessive)	2p13.3	254130	Liu (1998) Nat Genet 20, 31
Miyoshi muscular dystrophy 3	MMD3	*ANO5*	anoctamin 5	11p14.3	613319	Bolduc (2010) Am J Hum Genet 86, 213
Distal myoptahy, with anterior tibial onset	DMAT	*DYSF*			606768	Liu (1998) Nat Genet 20, 31
Distal myopathy 1 (Laing)	MPD1	*MYH7*	myosin, heavy chain 7, cardiac muscle, beta	14q11.2-q13	160500	Laing (1995) Am J Hum Genet 56
Tardive tibial muscular dystrophy (Udd myopathy)	TMD	*TTN*	titin	2q31	600334	Haravuori (1998) Am J Hum Genet. 62(3)
METABOLIC MYOPATHIES						
Glycogen storage disease Type II (Pompe disease)	GSD2	*GAA*	Glucosidase, alpha acid	17q25.2-q25.3	232300	Zhong (1991) Am J Hum Genet 49, 635
Glycogen storage disease Type V (McArdle disease)	GSD5	*PYGM*	Phosphorylase, glycogen (muscle)	11q12-q13.2	232600	Tsujino (1993) N Engl J Med 329, 241
Myoadenylate deaminase deficiency		*AMPD1*	adenosine monophosphate deaminase 1 (isoform M)	1p13	102770	Morisaki (1992) Proc Natl Acad Sci U S A 89, 6457
Carnitine palmitoyltransferase II deficiency		*CPT2*	Carnitine palmitoyltransferase 2	1p32	255110	Taroni (1993) Nat Genet 4, 314
OTHER MYOPATHIES						
Desmin-related myofibrillar myopathy	MFM	*DES*	desmin	2q35	601419	Goldfarb (1998) Nat Genet 19, 402
X-linked myopathy with postural muscle atrophy	XMPMA	*FHL1*	four and a half LIM domains 1	Xq26.3	300696	Windpassinger (2008) Am J Hum Genet 82, 88
Scapuloperoneal myopathy	XPMD				300695	Quinzii (2008) Am J Hum Genet 82, 208
Danon disease	(GSD2B)	*LAMP2*	Lysosomal-associated membrane protein 2	Xq24	300257	Nishino (2000) Nature 406, 906
Desmin-related myopathy with Mallory bodies	RSMD1	*SEPN1*	selenoprotein N, 1	1p36.13	602771	Ferreiro (2004) Ann. Neurol. 55
Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia	IBMPFD	*VCP*	Valosin-containing protein	9p13.3	167320	Watts (2004) Nat Genet 36, 377

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Disease	Abbreviation	Gene Symbol	Gene Name	Chromosome	MIM#	Key Reference
HyperCKemia						
Idiopathic hyperCKaemia		*CAV3*	caveolin 3	3p25	123320	Carbone (2000) Neurology 54
HyperCKemia		*CAPN3*	calpain 3, (p94)	15q15.1		Fanin (2009) Eur J Hum Genet 17, 598
HyperCKemia		*DYSF*	dysferlin, limb girdle muscular dystrophy 2B (autosomal recessive)	2p13.3		Nguyen (2007) Arch Neurol 64, 1176
Myoglobinuria, acute recurrent, autosomal recessive		*LPIN1*	lipin 1	2p25.1	268200	Zeharia (2008) Am J Hum Genet 83, 489
MYOTONIC SYNDROMES						
Myotonic dystrophy, type 1 (Steinert)	DM1	*DMPK*	dystrophia myotonica-protein kinase	19q13.3	160900	Brook (1992) Cell 68, 799
Myotonic dystrophy, type 2	DM2	*CNBP*	CCHC-type zinc finger, nucleic acid binding protein	3q21	602668	Liquori (2001) Science 293, 864
Proximal myotonic myopathy	PROMM	*CNBP*			602668	Liquori (2001) Science 293, 864
Rippling muscle disease	RM2	*CAV3*	caveolin 3	3p25	606072	Betz (2001) Nature Genet. 28
Congenital Generalized Lipodystrophy with Muscle Rippling	CGL4	*PTRF*	polymerase I and transcript release factor	17q21.2	613327	Rajab (2010) PLoS Genet 6:e1000874
Episodic ataxia/myokymia syndrome	EA1	*KCNA1*	potassium voltage-gated channel, Shaker-related subfamily, member 1	12p13.32	160120	Browne (1994) Nat Genet 8, 136
CONGENITAL MYASTHENIC SYNDROMES						
Multiple pterygium syndrome, Escobar variant		*CHRNG*	Cholinergic receptor, nicotinic, gamma	2q33-q34	265000	Morgan (2006) Am J Hum Genet 79, 390
Multiple pterygium syndrome, lethal type		*CHRNG*			253290	Hoffmann (2006) Am J Hum Genet 79, 303
Paramyotonia congenita of Von Eulenburg	PMC	*SCN4A*	Sodium channel, voltage gated, type IV, alpha polypeptide	17q23-q25.3	168300	McClatchey (1992) Nat Genet 2, 148
Hyperkalemic periodic paralysis, type 2	HYPP	*SCN4A*			170500	Rojas (1991) Nature 354, 387
Hypokalemic periodic paralysis, type 2	HOKPP2	*SCN4A*			613345	Bulman (1999) Neurology 53, 1932
Myotonia, potassium-aggravated		*SCN4A*			608390	Lerche (1993) J Physiol 470, 13
Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency	CMS1D	*RAPSN*	Receptor-associated protein of the synapse, 43kD (rapsyn)	11p11.2-p11.1	608931	Ohno (2002) Am J Hum Genet 70, 875
ION CHANNEL MUSCLE DISEASES						
Myotonia congenita, dominant		*CLCN1*	Chloride channel 1, skeletal muscle	7q34	160800	Koch (1992) Science 257, 797
Myotonia congenita, recessive					255700	
Brugada syndrome 1		*SCN5A*	Sodium channel, voltage gated, type V, alpha polypeptide	3p22.2	601144	Chen (1998) Nature 392, 293
Long QT syndrome-3					603830	Wang (1995) Hum Mol Genet 4, 1603
Long QT syndrome-1		*KCNQ1*	Potassium voltage gated channel, KQT-like subfamily, member 1 (KVLQT1)	11p15.5	192500	Wang (1996) Nat Genet 12, 17
Long QT syndrome-2		*KCNH2*	Potassium voltage-gated channel, subfamily H, member 2 (HERG)	7q35-q36	613688	Curran (1995) Cell 80, 795
SPINAL MUSCULAR ATROPHIES AND MOTOR NEURON DISEASES						
Spinal muscular atrophy, type I	SMA1	*SMN1*	survival of motor neuron 1, telomeric	5q13.2	253300	Lefebvre (1995) Cell 80, 155
Spinal muscular atrophy, type II	SMA2	*SMN1*			253550	
Spinal muscular atrophy, type III	SMA3	*SMN1*			253400	
Spinal muscular atrophy, type IV	SMA4	*SMN1*			271150	
Spinal muscular atrophy, proximal adult, autosomal dominant		*VAPB*	VAMP (vesicle-associated membrane protein)-associated protein B and C	20q13.33	182980	Nishimura (2004) Am J Hum Genet 75, 822
Spinal muscular atrophy with respiratory distress	SMARD1	*IGHMBP2*	immunoglobulin mu binding protein 2	11q13.3	604320	Grohmann (2001) Nat Genet 29, 75
Early onset myopathy, areflexia, respiratory distress and dysphagia	EMARDD	*MEGF10*	Multiple epidermal growth factor-like domains 10	5q32.2	614399	Logan (2011) Nat Genet 43, 1189
Spinal and bulbar muscular atrophy, X-linked 1	SMAX1	*AR*	androgen receptor (dihydrotestosterone receptor; testicular feminization; spinal and bulbar muscular atrophy; Kennedy disease)	Xq12	313200	LaSpada (1991) Nature 352, 77
Amyotrophic lateral sclerosis 1	ALS1	*SOD1*	superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult))	21q22.11	105400	Rosen (1993) Nature 362, 59
Amyotrophic lateral sclerosis 2, juvenile	ALS2	*ALS2*	Amyotrophic lateral sclerosis 2 (juvenile)	2q33.1	205100	Hadano (2001) Nat Genet 29, 166
Primary lateral sclerosis, juvenile	PLSJ				606353	Yang (2001) Nat Genet 29, 160
Spastic paralysis, infantile onset ascending	IAHSP				607225	Eymard-Pierre (2002) Am J Hum Genet 71, 518
Amyotrophic lateral sclerosis 8	ALS8	*VAPB*			608627	Nishimura (2004) Am J Hum Genet 75, 822
Amyotrophic lateral sclerosis 14 with or without frontotemporal dementia	ALS14	*VCP*	Valosin-containing protein	9p13.3	613954	Johnson (2010) Neuron 68, 857
HEREDITARY CARDIOMYOPATHIES						
Cardiomyopathy, familial hypertrophic	CMH	*CAV3*	caveolin 3	3p25	192600	
Cardiomyopathy, familial hypertrophic, 1	CMH1	*MYH7*	myosin, heavy chain 7, cardiac muscle, beta	14q11.2-q13	192600	Geisterfer-Lowrance (1990) Cell 62, 999
Cardiomyopathy, dilated, 1A	CMD1A	*LMNA*	lamin A/C	1q22	115200	Fatkin (1999) N Engl J Med 341, 1715
Cardiomyopathy, dilated, 1C	CMD1C	*LDB3*	LIM domain binding 3	10q22.3-q23.2	601493	Vatta (2003) J Am Coll Cardiol 42, 2014
Cardiomyopathy, dilated, 1E	CMD1E	*SCN5A*	Sodium channel, voltage gated, type V, alpha polypeptide	3p21	601154	Olson (2005) JAMA 293, 447
Cardiomyopathy, dilated, 1G	CMD1G	*TTN*	titin	2q31	604145	Satoh (1999) BNRC 262, 411
Dilated cardiomyopathy, 1I	CMD1I	*DES*	desmin	2q35	601419	Goldfarb (1998) Nat Genet 19, 402
Cardiomyopathy, dilated, 3B (X-linked)	CMD3B, XLCM	*DMD*	dystrophin (muscular dystrophy, Duchenne and Becker types)	Xp21.2	300376	
HEREDITARY MOTOR AND SENSORY NEUROPATHIES						
Charcot-Marie-Tooth disease, type 1A	CMT1A	*PMP22*	peripheral myelin protein 22	17p12	118220	Lupski (1991) Cell 66, 219
Charcot-Marie-Tooth disease, dominant intermediate B	CMTDIB	*DNM2*	dynamin 2	19p13.2	606482	Zuchner (2005) Nat Genet 37, 289
Hereditary neuropathy with pressure palsies	HNPP	*PMP22*			162500	Chance (1993) Cell. 15, 72
Dejerine-Sottas syndrome		*PMP22*			145900	Roa (1993) Nat Genet. 5, 3
Charcot-Marie-Tooth disease, axonal, type 2B1	CMT2B1	*LMNA*	lamin A/C	1q22	159001	Muchir (2000) Hum Mol Genet 9, 1453
Charcot-Marie-Tooth disease, type 2A2	CMT2A2	*MFN2*	mitofusin 2	1p36.22	609260	Zuchner (2004) Nat Genet 36, 449
Hereditary motor and sensory neuropathy VI	HMSN6	*MFN2*			601152	Zuchner (2006) Ann Neurol 59, 276
Charcot-Marie-Tooth neuropathy, X-linked dominant 1	CMTX1	*GJB1*	gap junction protein, beta 1, 32kDa	Xq13.1	302800	Bergoffen (1993) Science 262, 2039
Charcot-Marie-Tooth disease, type 4A	CMT4A	*GDAP1*	ganglioside-induced differentiation-associated protein 1	8q21.11	214400	Baxter (2002) Nat Genet 30, 21
Charcot-Marie-Tooth disease, axonal, type 2K	CMT2K	*GDAP1*			607831	
Charcot-Marie-Tooth disease, recessive intermediate, A	CMTRIA	*GDAP1*			608340	
Charcot-Marie-Tooth disease, axonal, with vocal cord paresis		*GDAP1*			607706	
Giant axonal neuropathy 1	GAN1	*GAN*	Giant axonal neuropathy	16q24.1	256850	Bomont (2000) Nat Genet 26, 370
NEURODEGENERATIVE LYSOSOMAL STORAGE DISORDERS						
Ceroid lipofuscinosis, neuronal, type 1	CLN1	*PPT1*	palmitoyl-protein thioesterase 1	1p32	256730	Vesa (1995) Nature 376, 584
Ceroid lipofuscinosis, neuronal, type 2	CLN2	*TPP1*	tripeptidyl peptidase I (CLN2)	11p15	204500	Sleat (1997) Science 277, 1802
Ceroid lipofuscinosis, neuronal, type 10	CLN10	*CTSD*	cathepsin D (lysosomal aspartyl protease)	11p15.5	610127	Steinfeld (2006) Am J Hum Genet 78, 988
DISORDERS OF NEURONAL MIGRATION						
Polymicrogyria, bilateral frontoparietal	BFPP	*GPR56*	G protein-coupled receptor 56	16q12.2-q21	606854	Piao (2004) Science 303, 2033
Lissencephaly, X-linked 1	LISX1	*DCX*	Doublecortex; lissencephaly, X-linked (doublecortin)	Xq22.3-q23	300067	Ross (1997) Hum Mol Genet 6, 555
Lissencephaly 1	LIS1	*PAFAH1B1*	Platelet activating factor acetylhydrolase, isoform Ib, alpha subunit (45kD)	17p13.3	607432	Reiner (1993) Nature 364, 717

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Disease	Abbreviation	Gene Symbol	Gene Name	Chromosome	MIM#	Key Reference
HEREDITARY STROKE DISORDERS						
Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy	CADASIL	*NOTCH3*	Notch (drosophila) homologue 3	19p13.2-p13.1	125310	Joutel (1996) Nature 383, 707
MITOCHONDRIAL DISORDERS						
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 1	PEOA1	*POLG*	polymerase (DNA directed), gamma	15q24	157640	Van Goethem (2001) Nat Genet 28, 211
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive	PEOB	*POLG*			258450	Van Goethem (2001) Nat Genet 28, 211
Sensory ataxic neuropathy, dysarthria, and ophtalmoparesis	SANDO	*POLG*			607459	
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3	PEOA3	*C10ORF2 (TWINKLE)*	chromosome 10 open reading frame 2 (Twinkle protein, mitochondrial, Ataxin 8)	10q23.3-q24.3	609286	Spelbrink (2001) Nat Genet 28, 223
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5	PEOA5	*RRM2B*	ribonucleotide reductase M2 B (TP53 inducible)	8q23.1	613077	
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	MTDPS1	*TYMP*	Thymidine phosphorylase (endothelial cell growth factor 1 (platelet-derived) ECGF1	22q13.33	603041	Nishino (1999) Science 283, 689
Mitochondrial DNA depletion syndrome 2 (myopathic type)	MTDPS2	*TK2*	thymidine kinase 2, mitochondrial	16q22	609560	Saada (2001) Nat Genet 29, 342
Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	MTDPS3	*DGUOK*	deoxyguanosine kinase	2p13	251880	Mandel (2001) Nat Genet 29, 337
Mitochondrial DNA depletion syndrome 4A (Alpers type)	MTDPS4A	*POLG*			203700	Ferrari (2005) Brain 128, 723
Mitochondrial DNA depletion syndrome 4B (MNGIE type)	MTDPS4B	*POLG*			613662	
Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	MTDPS7	*C10ORF2 (TWINKLE)*				
Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	MTDPS8A	*RRM2B*	ribonucleotide reductase M2 B (TP53 inducible)	8q23.1	612075	Bourdon (2007) Nat Genet 39, 776
Mitochondrial DNA depletion syndrome 8B (MNGIE type)	MTDPS8B	*RRM2B*				
Pyruvate dehydrogenase E3-binding protein deficiency		*PDHX*	pyruvate dehydrogenase complex, component X	11p13	245349	Ling (1998) Hum Mol Genet 7, 501
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency		*SCO2*	SCO (cytochrome oxidase deficient, yeast) homologue 2	22q13.33	604377	Papadopoulou (1999) Nat Genet 23, 333
DISORDERS OF OPTIC NERVE AND RETINA						
Stargardt disease 1	STGD1	*ABCA4*	ATP-binding cassette, sub-family A (ABC1), member 4 (Stargardt disease 1, ABCR)	1p22.1-p21	248200	Allikmets (1997) Nat Genet 15, 236
Stargardt disease 3	STGD3	*ELOVL4*	Elongation of very long chain fatty acids-like 4	6q14	600110	Zhang (2001) Nat Genet 27, 89
Cone-rod dystrophy 2	CORD2	*CRX*	Cone-rod homeobox	19q13.3	120970	Freund (1997) Cell 91, 543
Cone-rod dystrophy 3	CORD3	*ABCA4*			604116	Maugeri (2000) Am J Hum Genet 67, 960
Cone-rod dystrophy 6	CORD6	*GUCY2D*	Guanylate cyclase 2D, membrane (retina-specific)	17p13.1	601777	Kelsell (1998) Hum Mol Genet 7, 1179
Macular dystrophy, vitelliform	VMD	*BEST1*	Bestrophin 1 (VMD2)	11q13	153700	Petrukhin (1998) Nat Genet 19, 241
Macular dystrophy, vitelliform, adult-onset	AVMD	*BEST1*			608161	Krämer (2000) Eur J Hum Genet 8, 286
Macular dystrophy, vitelliform, adult-onset	AVMD	*PRPH2*	Peripherin 2 (retinal degeneration, slow) (RDS)	6p21.2-p12.3	608161	Wells (1993) Nat Genet 3, 213
Bestrophinopathy	ARB	*BEST1*			611809	Burgess (2008) Am J Hum Genet 82, 19
Vitreoretinochoroidopathy	VRCP	*BEST1*			193220	Burgess (2009) J Med Genet 46, 120
Macular degeneration, age-related 2	ARMD2	*ABCA4*			153800	Allikmets (1997) Science 277, 1805
Patterned dystrophy of retinal pigment epithelium		*PRPH2*			169150	Kohl (1997) J Med Genet 34, 620
Fundus albipunctatus		*PRPH2*			136880	Kajiwara (1993) Nat Genet 3, 208
Choroidal dystrophy, central areolar 2	CACD2	*PRPH2*			613105	Renner (2009) Am J Ophthalmol 147, 518
Leber congenital amaurosis 1	LCA1	*GUCY2D*	Guanylate cyclase 2D, membrane (retina-specific)	17p13.1	204000	Perrault (1996) Nat Genet 14, 461
Leber congenital amaurosis 2	LCA2	*RPE65*	Retinal pigment epithelium specific protein (65kD)	1p31.2-3	204100	Gu (1997) Nat Genet 17, 194
Leber congenital amaurosis 4	LCA4	*AIPL1*	Arylhydrocarbon-interacting protein-like-1	17p13.1	604393	Sohocki (2000) Nat Genet 24, 79
Leber congenital amaurosis 8	LCA8	*CRB1*	Crumbs, drosophila, homologue of, 1	1q31-q32.1	613835	den Hollander (2001) Am J Hum Genet 69, 198
Achromatopsia-2	ACHM2	*CNGA3*	Cyclic nucleotide gated channel alpha 3	2q11.2	216900	Kohl (1998) Nat Genet 19:257
Achromatopsia-3	ACHM3	*CNGB3*	Cyclic nucleotide gated channel, beta 3	8q21-q22	262300	Sundin (2000) Nat Genet 25, 289
Optic atrophy 1	OPA1	*OPA1*	Optic atrophy 1 (autosomal dominant)	3q28-q29	165500	Delettre (2000) Nat Genet 26, 207
Optic atrophy 1 and deafness		*OPA1*			125250	Yu-Wai-Man (2010) Brain 133, 771
Optic atrophy 3, autosomal dominant	OPA3	*OPA3*	Optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)	19q13.32	165300	Reynier (2004) J Med Genet 41, e110
Optic atrophy 7	OPA7	*TMEM126A*	Transmembrane protein 126A	11q14.1	612989	Hanein (2009) Am J Hum Genet 84, 493
Retinitis pigmentosa 1	RP1	*RP1*	Retinitis pigmentosa 1 (autosomal dominant)	8q11-q13	180100	Pierce (1999) Nat Genet 22, 248
Retinitis pigmentosa 2	RP2	*RP2*	Retinits pigmentosa 2 (X-linked recessive)	Xp11.4-p11.21	300757	Schwahn (1998) Nat Genet 19, 327
Retinitis pigmentosa 3	RP3	*RPGR*	Retinitis pigmentosa GTPase regulator	Xp11.4	300029	Meindl (1996) Nat Genet 13, 35
Retinitis pigmentosa 4	RP4	*RHO*	Rhodopsin	3q21-q24	180380	Dryja (1990) Nature 343, 364
Retinitis pigmentosa 7	RP7	*PRPH2*	Peripherin 2 (retinal degeneration, slow) (RDS)	6p21.2-p12.3	608133	Kajiwara (1991) Nature 354, 480
Retinitis pigmentosa 11	RP11	*PRPF31*	PRP31 pre-mRNA processing factor 31 homologue (yeast)	19q13.42	600138	Vithana (2001) Mol Cell 8, 375
Retinitis pigmentosa 12	RP12	*CRB1*			604210	den Hollander (1999) Nat Genet 23, 217
Retinitis pigmentosa 19	RP19	*ABCA4*			601718	Martinez-Mir (1998) Nat Genet 18, 11
Retinitis pigmentosa 39	RP39	*USH2A*	Usher syndrome 2A, (autosomal recessive, mild)	1q41	608400	
Retinitis pigmentosa 50	RP50	*BEST1*			613194	Davidson (2009) Am J Hum Genet 85, 581
Doyne Honeycomb retinal dystrophy (drusen, radial, autosomal dominant)	DHRD	*EFEMP1*	EGF-containing fibulin-like extracellular matrix protein 1	2p16	126600	Stone (1999) Nat Genet 22, 199
Basal laminar drusen		*CFH*		1q32	126700	Boon (2008) Am J Hum Genet 82, 516
Wolfram syndrome 1	WFS1	*WFS1*	Wolfram syndrome 1 (wolframin)	4p16	222300	Inoue (1998) Nat Genet 20, 143
Albinism, ocular, type I	OA1	*GPR143*	G protein-coupled receptor 143 (OA1)	Xp22.3	300500	Bassi (1995) Nat Genet 10, 13
Albinism, oculocutaneous, type IA	OCA1A	*TYR*	Tyrosinase	11q14.3	203100	Tomita (1989) Biochem Biophys Res Commun 164, 990
Albinism, oculocutaneous, type IB	OCA1B				606952	
Albinism, oculocutaneous, type II	OCA2	*OCA2*	Oculocutaneous albinism II	15q11.2-q12	611409	Rinchik (1993) Nature 361, 72
Retinoschisis 1, X-linked, juvenile	RS1	*RS1*	Retinoschisis (X linked, juvenile) 1	Xp22.2-p22.1	312700	Sauer (1997) Nat Genet 17, 164
Usher Syndrome, type IB	USH1B	*MYO7A*	Myosin VIIA	11q13.5	276900	Weil (1995) Nature 374, 60
Usher Syndrome, type IIA	USH2A	*USH2A*	Usher syndrome 2A, (autosomal recessive, mild)	1q41	276901	Eudy (1998) Science 280, 1753
Usher Syndrome, type IIC	USH2C	*GPR98*	G protein-coupled receptor 98	5q13	605472	Nakayama (2002) Ann Neurol 52, 654
Usher Syndrome, type IID	USH2D	*DFNB31*	Deafness, autosomal recessive 31 (WHRN)	9q32-q34	611383	Mburu (2003) Nat Genet 34, 421
Hypotrichosis, congenital, with juvenile macular dystrophy	HJMD	*CDH3*	Cadherin 3, type 1, P-cadherin (placental)	16q22.1	601553	Sprecher (2001) Nat Genet 29, 134
Sorsby fundus dystrophy	SFD	*TIMP3*	Tissue inhibitor of metalloproteinase 3	22q12.3	136900	Weber (1994) Nat Genet 8, 352

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Disease	Abbreviation	Gene Symbol	Gene Description	Nucleotide Positions (RCRS) of Mutation(s) bold: confirmed status in Mitomap	MIM#
MITOCHONDRIAL GENOME MUTATION CAUSED DISORDERS					
Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like episodes	MELAS	*MTTL1*	tRNA leucine (UUR)	**A3243G**, A3243T, G3244A, A3252G, **C3256T**, T3258C, T3271C, **T3291C**	540000
		MTTH	tRNA histidine	**G12147A**	
		MTTL2	tRNA leucine (CUN)	A12299C	
		MTTE	tRNA glutamic acid	A14693G	
		MTTF	tRNA phenylanine	**G583A**	
		MTTV	tRNA valine	G1642A	
		MTTQ	tRNA glutamine	**G4332A**	
		MTRNR2	16S ribosomal RNA	C3093G	
		MTND1	NADH dehydrogenase subunit 1	T3308C, G3376A, G3481A, **G3697A**, G3946A, T3949C	
		MTCO3	Cytochrome c oxidase subunit III	T9957C	
		MTND4	NADH dehydrogenase subunit 4	A11084G	
		MTND5	NADH dehydrogenase subunit 5	A12770G, A13045C, A13084T, **G13513A**, **A13514G**, A13849C	
		MTND6	NADH dehydrogenase subunit 6	G14453A	
		MTCYB	Cytochrome b	14787del4	
Myoclonic Epilepsy and Ragged Red Muscle Fibers	MERRF	*MTTK*	tRNA lysine	A8296G, **A8344G**, **T8356C**, G8361A, **G8363A**	545000
		MTTH	tRNA histidine	**G12147A**	
		MTTL1	tRNA leucine (UUR)	G3255A	
		MTTF	tRNA phenylanine	G611A	
MERRF/MELAS overlap disease	MERME	*MTTS1*	tRNA serine (UCN)	T7512C	
Myoclonic Epilepsy and Psychomotor Regression	MEPR	*MTTD*	tRNA aspartic acid	A7543G	
Progressive Encephalopathy	PEM	*MTTS1*	tRNA serine (UCN)	**C7471C**, T7512C	
		MTTG	tRNA glycine	**T10010C**	
		MTTL1	tRNA leucine (UUR)	T3271delT, C3287A	
		MTTT	tRNA threonine	G15915A	
		MTTW	tRNA tryptophan	G5540A	
		MTTE	tRNA glutamic acid	T14709C, G14740A	
		MTND1	NADH dehydrogenase subunit 1	G3481A, G3890A	
		MTND2	NADH dehydrogenase subunit 2	C5452T	
		MTCYB	Cytochrome b	G15242A	
		MTCO2	Cytochrome c oxidase subunit II	T7587C, G7859A	
		MTCO3	Cytochrome c oxidase subunit III	T9957C	
Epilepsy, Strokes, Optic atrophy, & Cognitive decline	ESOC	*MTND3*	NADH dehydrogenase subunit 3	**T10191C**	
Leigh Disease (Maternally Inherited Leigh Syndrome)	LD (MILS)	*MTND1*	NADH dehydrogenase subunit 1	G3688A	256000
		MTND2	NADH dehydrogenase subunit 2	T4681C	
		MTND3	NADH dehydrogenase subunit 3	**T10158C**, **T10191C**, **G10197A**	
		MTND4	NADH dehydrogenase subunit 4	**C11777A**	
		MTND5	NADH dehydrogenase subunit 5	**T12706C**, A13045C, A13084T, **G13513A**, **A13514G**	
		MTND6	NADH dehydrogenase subunit 6	**G14459A**, **T14487C**, G14600A	
		MTATP6	ATP synthase F0 subunit 6	**T8993C**, **T8993G**, **T9176C**, T9176G, **T9185C**, T9191C	
		MTCO3	Cytochrome c oxidase subunit III	C9537insC	
		MTTW	tRNA tryptophan	**A5537insT**	
		MTTV	tRNA valine	C1624T, G1644T	
Kearns-Sayre Syndrome	KSS	-	-	**various deletions**	530000
		MTTL2	tRNA leucine (CUN)	**G12315A**	
		MTTL1	tRNA leucine (UUR)	G3249A, G3255A	
Pearson Syndrome		-	-	**various deletions**	557000
Leber Hereditary Optic Neuropathy	LHON	*MTTQ*	tRNA glutamine	A4381G	535000
		MTTL1	tRNA leucine (UUR)	C3275A	
		MTTT	tRNA threonine	A15951G	
		MTTM	tRNA methionine	A4435G	
		MTND1	NADH Dehydrogenase subunit 1	G3376A, **G3460A**, **G3635A**, **G3700A**, **G3733A**, T4160C, **C4171A**	
		MTND2	NADH Dehydrogenase subunit 2	C4640A, G5244A	
		MTND3	NADH Dehydrogenase subunit 3	T10237C	
		MTND4	NADH dehydrogenase subunit 4	G11696A, **G11778A**, C11874A	
		MTND4L	NADH dehydrogenase subunit 4L	A10543G, T10591G, **T10663C**	
		MTND5	NADH dehydrogenase subunit 5	T12782G, C12848T, A13045C, G13051A, A13379C, A13528G, G13730A	
		MTND6	NADH dehydrogenase subunit 6	G13730A, G14279A, T14325C, C14482A, **C14482G**, **T14484C**, A14495G, T14498C, **C14568T**, A14596T, **A14495G**	
		MTCO1	Cytochrome c oxidase subunit I	G6261A, G7444A	
		MTCO2	Cytochrome c oxidase subunit II	C7623T	
		MTCO3	Cytochrome c oxidase subunit II	A9660C, G9738T, G9804A	
		MTATP6	ATP synthase F0 subunit 6	A8836G, A9016G, T9101C	
		MTCYB	Cytochrome b	T15674C	
Leber's hereditary optic neuropathy and Dystonia	LDYT	*MTND6*	NADH dehydrogenase subunit 6	**G14459A**	500001
		MTATP6	ATP synthase F0 subunit 6	G8950A	

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Disease	Abbreviation	Gene Symbol	Gene Description	Nucleotide Positions (RCRS) of Mutation(s) bold: confirmed status in Mitomap	MIM#
MITOCHONDRIAL GENOME MUTATION CAUSED DISORDERS					
Mitochondrial Myopathy	MM	*MTTA*	tRNA alanine	**G5650A**	
		MTTN	tRNA asparagine	T5692C, G5698A, **G5703A**	
		MTTS1	tRNA serine (UCN)	A7472C, T7480G, **G7497A**	
		MTTE	tRNA glutamic acid	A14687G, **T14709C**	
		MTTF	tRNA phenylanine	**G583A**, T618C	
		MTTL1	tRNA leucine (UUR)	G3242A, **A3243G**, A3243T, T3250C, A3251G, C3254G, T3258C, A3280G, A3288G, **A3302G**, A4267G	
		MTTL2	tRNA leucine (CUN)	A12320G	
		MTCO2	Cytochrome c oxidase subunit II	T7671A	
		MTCO3	Cytochrome c oxidase subunit III	del9486:9502, G9379A, T9789C	
Lethal Infantile Mitochondrial Myopathy	LIMM	*MTTT*	tRNA threonine	A15923G, A15924G	551000
Maternally Inherited Diabetes and Deafness	MIDD	*MTTL1*	tRNA leucine (UUR)	**A3243G**	520000
		MTND1	NADH dehydrogenase subunit 1	G3421A	
		MTATP8	ATP synthase F0 subunit 8	A8381G	
Diabetes insipidus and mellitus with Optic Atrophy and Deafness (Wolfram Syndrome, mitochondrial form)	DIDMOAD	-	-	**deletion**	598500
Diabetes Mellitus	DM	*MTTL1*	tRNA leucine (UUR)	T3264C, T3271C	
		MTND4	NADH Dehydrogenase subunit 4	A12026G	
		MTND6	NADH Dehydrogenase subunit 6	T14577C	
Non-Insulin Dependent Diabetes Mellitus	NIDDM	*MTND1*	NADH Dehydrogenase subunit 1	C3310T	
Diabetes Mellitus & Deafness	DMDF	*MTTL1*	tRNA leucine (UUR)	**A3243G**	590050
		MTTE	tRNA glutamic acid	**T14709C**	
		MTTK	tRNA lysine	A8296G	
		MTTS1	tRNA serine (UCN)	A7472C	
		MTTS2	tRNA serine (AGY)	C12258A	
		MTND1	NADH Dehydrogenase subunit 1	A3398G	
Maternally inherited deafness	DEAF	*MTRNR1*	12S ribosomal RNA	T990C, A1116G, **C1494T**, A1517C, **A1555G**	520000
		MTCO1	Cytochrome c oxidase subunit I	A7443G, G7444A, **A7445C**	
		MTND5	NADH Dehydrogenase subunit 5	T12338C	
		MTTW	tRNA tryptophan	G5540A	
		MTTA	tRNA alanine	T5628C, T5655C	
		MTTC	tRNA cysteine	G5783A, T5802C	
		MTTS1	tRNA Ser (UCN)	**A7445C**	
		MTTK	tRNA lysine	**G8363A**	
		MTTH	tRNA histidine	G12183A	
		MTTF	tRNA phenylalanine	G622A, A636G, T642C	
		MTTL1	tRNA leucine (UUR)	**T3291C**	
Sensorineural Hearing Loss	SNHL	*MTCO1*	Cytochrome c oxidase subunit I	G7444A, **A7445G**	
		MTCO2	Cytochrome c oxidase subunit II	A8108G	
		MTND6	NADH dehydrogenase subunit 6	C14340T	
		MTCYB	Cytochrome b	G15699C	
		MTRNR1	12S ribosomal RNA	T1095C	
		MTTC	tRNA cysteine	G5780A	
		MTTS1	tRNA serine (UCN)	**A7445G**, A7445T, T7510C, **T7511C**	
		MTTS2	tRNA serine (AGY)	C12258A	
		MTTL1	tRNA leucine (UUR)	**A3243G**	
(Chronic) Progressive External Ophthalmoplegia	(C)PEO	*MTTN*	tRNA asparagine	T5692C, G5698A, **G5703A**	
		MTTS2	tRNA serine (AGY)	G12276A	
		MTTL2	tRNA leucine (CUN)	G12294A, T12311C, **G12315A**, G12316A	
		MTTL1	tRNA leucine (UUR)	**A3243G**, T3250C, C3254T	
		MTTI	tRNA isoleucine	T4274C, T4285C, **G4298A**, **G4308A**, G4309A	
		MTTA	tRNA alanine	T5628C	
		MTND4	NADH dehydrogenase subunit 4	T11232C	
Maternal Myopathy and Cardiomyopathy	MMC	*MTTL1*	tRNA leucine (UUR)	**A3260G**, **C3303T**	
Maternally Inherited Hypertrophic Cardiomyopathy	MHCM	*MTTI*	tRNA isoleucine	A4295G	
		MTTG	tRNA glycine	T9997C	
Maternally Inherited Cardiomyopathy	MICM	*MTTI*	tRNA isoleucine	**A4300G**	
		MTTK	tRNA lysine	A8348G, **G8363A**	
		MTTH	tRNA histidine	G12192A	
Fatal Infantile Cardiomyopathy Plus, a MELAS-associated cardiomyopathy	FICP	*MTTI*	tRNA isoleucine	A4269G, A4317G	
Mitochondrial Encephalocardiomyopathy		*MTTI*	tRNA isoleucine	C4320T	
Alzheimer's Disease	AD	*MTND2*	NADH dehydrogenase subunit 2	G5460T	502500
		MTCO3	Cytochrome c oxidase subunit III	T9861C	
Alzheimer's Disease and Parkinsons's Disease	ADPD	*MTND1*	NADH dehydrogenase subunit 1	A3397G	556500
		MTRNR2	16S ribosomal RNA	G3196A	
Dementia and Chorea	DEMCHO	*MTTW*	tRNA tryptophan	G5549A	590095
Familial Bilateral Striatal Necrosis	FBSN	*MTATP6*	ATP synthase F0 subunit 6	**T9176C**	500003
Neurogenic muscle weakness, Ataxia, and Retinitis Pigmentosa	NARP	*MTATP6*	ATP synthase F0 subunit 6	**T8993G**, **T8993C**, **T9185C**	551500
Seizures/Lacticacidemia		*MTATP6*	ATP synthase F0 subunit 6	9205del2	
Tubulointerstitial nephritis		*MTTF*	tRNA phenylanine	A608G	551200
Ataxia, Myoclonus and Deafness	AMDF	*MTTV*	tRNA valine	**G1606A**	159800
		MTTS1	tRNA serine (UCN)	**C7471CC**	