MutationsCodingControl lt MITOMAP lt Foswiki

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
114	MT-CR	BD-associated	C114T	C-T	noncoding	+	-	Reported	188	1
150	MT-CR	Longevity / Cervical Carcinoma / HPV infection risk	C150T	C-T	noncoding	+	+	Conflicting reports	5584	8
195	MT-CR	BD-associated / melanoma pts	T195C	T-C	noncoding	+	-	Reported	8308	3
302	MT-CR	Higher in melanoma patient group	A302ACC	A-ACC	noncoding	.	.	Reported	74	1
309	MT-CR	AD-weakly associated	C309CC	C-CC	noncoding	.	.	Reported	308	1
310	MT-CR	Melanoma patients	T310TC	T-TC	noncoding	.	.	Reported	0	1
3308	MT-ND1	MELAS / DEAF enhancer / hypertension / LVNC / putative LHON	T3308C	T-C	M-T	-	+	P.M.-possibly synergistic	326	14
3308	MT-ND1	Sudden Infant Death	T3308G	T-G	M-X	+	+	Reported	6	1
3310	MT-ND1	Diabetes / HCM	C3310T	C-T	P-S	+	+	Reported	11	3
3316	MT-ND1	Diabetes / LHON / PEO	G3316A	G-A	A-T	+	-	Unclear	381	18

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
3335	MT-ND1	LHON (putative)	T3335C	T-C	I-T	+	-	Reported	41	1
3337	MT-ND1	Cardiomyopathy	G3337A	G-A	V-M	+	-	Possibly synergistic	70	1
3340	MT-ND1	Encephaloneuromyopathy	C3340T	C-T	P-S	+	-	Reported	3	1
3376	MT-ND1	LHON MELAS overlap	G3376A	G-A	E-K	+	+	Cfrm	0	3
3380	MT-ND1	MELAS	G3380A	G-A	R-Q	-	+	Reported	3	1
3388	MT-ND1	Materally Inherited Nonsyndromic Deafness	C3388A	C-A	L-M	.	.	Reported	16	1
3391	MT-ND1	LHON (putative)	G3391A	G-A	G-S	+	-	Reported	45	1
3394	MT-ND1	LHON / Diabetes / CPTdeficiency / high altitude adaptation	T3394C	T-C	Y-H	+	-	Reported / Unclear	580	27
3395	MT-ND1	LHON / HCM with hearing loss	A3395G	A-G	Y-C	+	+	Reported	20	3
3396	MT-ND1	NSHL / MIDD	T3396C	T-C	syn	+	-	Reported / Unclear	308	2

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
3397	MT-ND1	ADPD / Possibly LVNC-cardiomyopathy associated	A3397G	A-G	M-V	+	-	Reported	120	10
3398	MT-ND1	DMDF+HCM / GDM / possibly LVNC cardiomyopathy-associated	T3398C	T-C	M-T	+	-	Reported	166	5
3399	MT-ND1	Gestational Diabetes (GDM)	A3399T	A-T	M-I	+	-	Reported	12	1
3407	MT-ND1	HCM / Muscle involvement	G3407A	G-A	R-H	+	-	Conflicting reports	1	3
3418	MT-ND1	AMegL	A3418G	A-G	N-D	+	-	Reported	1	1
3421	MT-ND1	MIDD	G3421A	G-A	V-I	+	-	Reported	68	1
3460	MT-ND1	LHON	G3460A	G-A	A-T	+	+	Cfrm	20	138
3472	MT-ND1	LHON	T3472C	T-C	F-L	+	-	Reported	5	2
3481	MT-ND1	MELAS	G3481A	G-A	E-K	-	+	Reported	0	2
3481	MT-ND1	Progressive Encephalomyopathy	G3481A	G-A	E-K	-	+	Reported	0	1
3488	MT-ND1	LHON (putative)	T3488C	T-C	L-P	+	-	Reported	1	1
3496	MT-ND1	LHON	G3496T	G-T	A-S	+	-	Reported / Secondary	11	2

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
3497	MT-ND1	LHON	C3497T	C-T	A-V	+	-	Reported / Secondary	129	3
3551	MT-ND1	LHON (putative)	C3551T	C-T	A-V	+	-	Reported	0	1
3632	MT-ND1	LHON (putative)	C3632T	C-T	S-F	+	-	Reported	0	1
3634	MT-ND1	LHON	A3634G	A-G	S-G	+	-	Reported	0	1
3635	MT-ND1	LHON	G3635A	G-A	S-N	+	-	Cfrm	9	8
3644	MT-ND1	BD-associated	T3644C	T-C	V-A	.	.	Reported	189	3
3688	MT-ND1	Leigh Syndrome	G3688A	G-A	A-T	+	-	Reported	0	2
3697	MT-ND1	MELAS / LS / LDYT	G3697A	G-A	G-S	+	+	Cfrm	0	10
3700	MT-ND1	LHON	G3700A	G-A	A-T	+	-	Cfrm	3	2
3713	MT-ND1	LHON (putative)	T3713C	T-C	V-A	+	-	Reported	0	1
3733	MT-ND1	LHON	G3733A	G-A	E-K	+	+	Cfrm	2	6
3733	MT-ND1	LHON	G3733C	G-C	E-Q	-	+	Reported	0	1
3736	MT-ND1	LHON	G3736A	G-A	V-I	.	.	Reported	70	1
3745	MT-ND1	Possible adaptive high altitude variant	G3745A	G-A	A-T	.	.	Reported	95	1
3769	MT-ND1	LHON (putative)	C3769G	C-G	L-V	+	-	Reported	0	1
3781	MT-ND1	LHON (putative)	T3781C	T-C	S-P	+	-	Reported	0	1
3796	MT-ND1	Adult-Onset Dystonia	A3796G	A-G	T-A	-	+	Reported	187	3

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
3833	MT-ND1	PEG	T3833A	T-A	L-Q	+	-	Reported	0	1
3866	MT-ND1	LHON+limb claudication	T3866C	T-C	I-T	.	.	Reported	122	2
3890	MT-ND1	Progressive Encephalomyopathy / LS / Optic Atrophy	G3890A	G-A	R-Q	-	+	Cfrm	1	4
3902	MT-ND1	EXIT+myalgia / severe LA+cardiac / 3-MGA aciduria	3902_3908invACCTTGC	inversion	DLA-GKV	-	+	Cfrm	0	3
3919	MT-ND1	LHON (putative)	T3919C	T-C	S-P	+	-	Reported	0	1
3946	MT-ND1	MELAS	G3946A	G-A	E-K	+	+	Reported	2	6
3949	MT-ND1	MELAS	T3949C	T-C	Y-H	-	+	Reported	1	6
3958	MT-ND1	LHON (putative)	G3958A	G-A	G-S	+	-	Reported	0	1
3959	MT-ND1	MELAS	G3959A	G-A	G-D	.	.	Reported	0	1
3995	MT-ND1	MELAS	A3995G	A-G	N-S	.	.	Reported	15	1
4081	MT-ND1	LHON (putative)	T4081C	T-C	F-L	+	-	Reported	0	1
4123	MT-ND1	LHON (putative)	A4123T	A-T	I-F	+	-	Reported	0	1
4132	MT-ND1	NAION-associated	G4132A	G-A	A-T	+	-	Reported	7	1
4136	MT-ND1	LHON	A4136G	A-G	Y-C	+	-	Possibly synergistic	54	10

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
4142	MT-ND1	Developmental delay, seizure, hypotonia	G4142A	G-A	R-Q	-	+	Reported	0	1
4160	MT-ND1	LHON	T4160C	T-C	L-P	+	-	Reported	1	10
4163	MT-ND1	LHON (putative)	T4163C	T-C	M-T	+	-	Reported	0	1
4171	MT-ND1	LHON	C4171A	C-A	L-M	+	+	Cfrm	2	7
4216	MT-ND1	LHON / Insulin Resistance /possible adaptive high altitude variant	T4216C	T-C	Y-H	+	-	P.M. - haplogroup J / T marker	4267	37
4633	MT-ND2	LHON candidate	C4633G	C-G	A-G	+	-	Reported	0	1
4640	MT-ND2	LHON	C4640A	C-A	I-M	+	-	Reported	117	4
4648	MT-ND2	PEG	T4648C	T-C	F-S	+	-	Reported	1	1
4659	MT-ND2	possible PD risk factor	G4659A	G-A	A-T	+	-	Reported	57	1
4681	MT-ND2	Leigh Syndrome	T4681C	T-C	L-P	-	+	Reported	1	2
4769	MT-ND2	SZ-associated	A4769A	A-A	syn	+	-	Reported	0	2
4833	MT-ND2	Diabetes helper mutation; AD, PD	A4833G	A-G	T-A	+	-	Reported; haplogroup G marker	343	2
4852	MT-ND2	LHON	T4852A	T-A	L-Q	+	-	Reported	0	1
4883	MT-ND2	Glaucoma	C4883T	C-T	syn	+	-	Conflicting reports	2068	2

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
4917	MT-ND2	LHON / Insulin Resistance / AMD / NRTI-PN	A4917G	A-G	N-D	+	-	Reported; haplogroup T marker	2026	27
5001	MT-ND2	Developmental delay, seizure, cardiomyopathy, lactic acidosis	A5001AA	A-AA	frameshift	-	+	Reported	0	2
5134	MT-ND2	Exercise intolerance (EXIT)	AA5134d	AAA-A	frameshift	.	.	Reported	0	5
5178	MT-ND2	Longevity; Extraversion MI / AMS protection; blood iron metabolism	C5178A	C-A	L-M	+	-	Reported; haplogroup D marker	2053	18
5244	MT-ND2	LHON	G5244A	G-A	G-S	-	+	Reported	0	7
5452	MT-ND2	Progressive Encephalomyopathy	C5452T	C-T	T-M	+	-	Reported	13	1
5460	MT-ND2	AD / PD	G5460A	G-A	A-T	+	+	P.M.	2654	8
5460	MT-ND2	AD	G5460T	G-T	A-S	+	+	Reported	0	5
5911	MT-CO1	Prostate Cancer	C5911T	C-T	A-V	+	-	Reported	192	1
5913	MT-CO1	Prostate Cancer / hypertension	G5913A	G-A	D-N	+	-	Reported	420	3

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
5920	MT-CO1	Myoglobinuria / EXIT	G5920A	G-A	W-Ter	-	+	Reported	0	4
5935	MT-CO1	Prostate Cancer	A5935G	A-G	N-S	+	-	Reported	1	1
5973	MT-CO1	Prostate Cancer	G5973A	G-A	A-T	+	-	Reported	8	1
6020	MT-CO1	Motor Neuron Disease	CGAGC6020d	CGAGC-del	AELGQ-AGPATer	-	+	Reported	0	1
6081	MT-CO1	Prostate Cancer	G6081A	G-A	A-T	+	-	Reported	1	1
6150	MT-CO1	Prostate Cancer / enriched in POAG cohort	G6150A	G-A	V-I	+	-	Reported	204	2
6253	MT-CO1	Prostate Cancer / enriched in POAG cohort	T6253C	T-C	M-T	+	-	Reported	430	3
6261	MT-CO1	Prostate Cancer / LHON	G6261A	G-A	A-T	+	-	Reported	307	3
6267	MT-CO1	Prostate Cancer	G6267A	G-A	A-T	+	-	Reported	66	1
6285	MT-CO1	Prostate Cancer	G6285A	G-A	V-I	+	-	Reported	114	1
6328	MT-CO1	EXIT (Exercise Intolerance)	C6328T	C-T	S-F	+	-	Reported	0	2
6340	MT-CO1	Prostate Cancer	C6340T	C-T	T-I	+	-	Reported	75	2
6480	MT-CO1	Prostate Cancer / enriched in POAG cohort	G6480A	G-A	V-I	+	-	Reported	131	4
6489	MT-CO1	Therapy-Resistant Epilepsy	C6489A	C-A	L-I	-	+	Reported	70	2

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
6597	MT-CO1	MELAS-like syndrome	C6597A	C-A	Q-K	-	+	Reported	0	1
6663	MT-CO1	Prostate Cancer	A6663G	A-G	I-V	+	-	Reported	143	3
6698	MT-CO1	Myopathy	A6698del	A-del	K-fs-Ter	-	+	Reported	0	1
6708	MT-CO1	MM & Rhabdomyolysis	G6708A	G-A	G-Ter	-	+	Reported	0	1
6721	MT-CO1	Acquired Idiopathic Sideroblastic Anemia	T6721C	T-C	M-T	-	+	Reported	0	2
6742	MT-CO1	Acquired Idiopathic Sideroblastic Anemia	T6742C	T-C	I-T	-	+	Reported	0	2
6930	MT-CO1	Multisystem Disorder	G6930A	G-A	G-Ter	-	+	Reported	0	3
6955	MT-CO1	Mild EXIT and MR	G6955A	G-A	G-D	+	+	Reported	1	1
6962	MT-CO1	Possible helper variant for 15927A	G6962A	G-A	L-L	+	-	Reported	867	1
7023	MT-CO1	MELAS-like syndrome	G7023A	G-A	V-M	-	+	Reported	0	1
7041	MT-CO1	Prostate Cancer	G7041A	G-A	V-I	+	-	Reported	5	1
7080	MT-CO1	Prostate Cancer	T7080C	T-C	F-L	+	-	Reported	40	1
7083	MT-CO1	Prostate Cancer	A7083G	A-G	I-V	+	-	Reported	15	1
7158	MT-CO1	Prostate Cancer	A7158G	A-G	I-V	+	-	Reported	29	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
7305	MT-CO1	Prostate Cancer	A7305C	A-C	M-L	+	-	Reported	0	1
7402	MT-CO1	Isolated complex IV deficiency	C7402del	C-del	frameshift: P-HPTTHSKNPYTX	-	+	Reported	0	1
7443	MT-CO1	DEAF	A7443G	A-G	Ter-G	+	-	Reported	1	4
7444	MT-CO1	LHON / SNHL / DEAF	G7444A	G-A	Ter-K	+	-	Reported	146	24
7445	MT-CO1	DEAF	A7445C	A-C	Ter-S	+	-	Reported	10	5
7445	MT-CO1	SNHL	A7445G	A-G	Ter-Ter	+	+	Cfrm	1	28
7587	MT-CO2	Mitochondrial Encephalomyopathy	T7587C	T-C	M-T	-	+	Reported	0	2
7598	MT-CO2	Possible LHON helper variant	G7598A	G-A	A-T	-	+	Reported	580	2
7623	MT-CO2	LHON	C7623T	C-T	T-I	+	-	Reported	0	1
7637	MT-CO2	PD risk factor	G7637A	G-A	E-K	-	+	Reported	2	1
7671	MT-CO2	MM	T7671A	T-A	M-K	-	+	Reported	0	2
7697	MT-CO2	Possible HCM susceptibility	G7697A	G-A	V-I	+	-	Reported	242	3
7706	MT-CO2	Alpers-Huttenlocher-like	G7706A	G-A	A41T		+	Reported	8	1
7859	MT-CO2	Progressive Encephalomyopathy	G7859A	G-A	D-N	+	-	Reported	132	1
7868	MT-CO2	LHON	C7868T	C-T	L-F	+	-	Possibly synergistic	16	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
7877	MT-CO2	PEG glaucoma	A7877C	A-C	K-Q	+	-	Reported	0	1
7896	MT-CO2	Multisystem Disorder	G7896A	G-A	W-Ter	-	+	Reported	0	1
7970	MT-CO2	Encephalopathy	G7970T	G-T	E-Ter	-	+	Reported	0	1
7989	MT-CO2	Rhabdomyolysis	T7989C	T-C	L-P	-	+	Reported	0	2
8010	MT-CO2	Developmental delay, ataxia, seizure, hypotonia, lactic acidosis	T8010C	T-C	V-A	-	+	Reported	2	1
8021	MT-CO2	Asthenozoospermia	A8021G	A-G	I-V	+	-	Reported	4	1
8042	MT-CO2	Lactic Acidosis	8042delAT	AT-del	M-Ter	-	+	Reported	0	1
8078	MT-CO2	DEAF	G8078A	G-A	V-I	+	-	Reported	26	2
8108	MT-CO2	SNHL	A8108G	A-G	I-V	+	-	Reported	64	1
8156	MT-CO2	Multi-system mitochondrial disorder	G8156del	G-del	frameshift	-	+	Reported	0	1
8249	MT-CO2	Mitochondrial myopathy	G8249A	G-A	G-Ter	+	-	Reported	0	2
8381	MT-ATP8	MIDD / LVNC cardiomyopathy-assoc.	A8381G	A-G	T-A	+	-	Reported	9	2
8393	MT-ATP8	Reversible brain pseudoatrophy	C8393T	C-T	P-S	-	+	Reported	154	2

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
8403	MT-ATP8	Episodic weakness and progressive neuropathy	T8403C	T-C	I-T	+	-	Reported	1	1
8411	MT-ATP8	Severe mitochondrial disorder	A8411G	A-G	M-V	+	-	Reported	2	1
8414	MT-ATP8	Longevity	C8414T	C-T	L-F	+	-	Reported	1711	1
8481	MT-ATP8	Tetralogy of Fallot patient	C8481T	C-T	P-L	+	-	Reported	3	1
8519	MT-ATP8	Susceptibility to bullous pemphigoid	G8519A	G-A	E-K	+	-	Reported	103	1
8527	MT-ATP8 / 6	Neuromuscular disorder, possible helper mutation	A8527G	A-G	ATP8:K-K; ATP6:M(start)-V	+	-	Reported	145	1
8528	MT-ATP8 / 6	Infantile cardiomyopathy	T8528C	T-C	ATP8:W-R; ATP6:M(start)-T	+	+	Cfrm	0	3
8529	MT-ATP8 / 6	Apical HCM	G8529A	G-A	ATP8:W-X; ATP6:M-M	+	-	Reported	0	1
8558	MT-ATP8 / 6	Possibly LVNC cardiomyopathy-associated	C8558T	C-T	ATP8:P-S; ATP6:A-V	+	-	Reported	12	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
8561	MT-ATP8 / 6	Ataxia w neuropathy, DM, SNHL, and hypogonadism	C8561G	C-G	ATP8:P-A; ATP6:P-R	+	+	Reported	0	1
8611	MT-ATP6	Ataxia, microcephaly, developmental delay, intellectual disability	C8611CC	C-CC	frameshift	-	+	Reported	0	1
8618	MT-ATP6	NARP	T8618TT	T-TT	truncated protein	-	+	Reported	0	1
8668	MT-ATP6	LHON	T8668C	T-C	W-R	+	-	Reported	32	1
8719	MT-ATP6	Suspected mito disease	G8719A	G-A	G-Ter	-	+	Reported	0	1
8741	MT-ATP6	MILS protective factor	T8741G	T-G	L-R	-	+	Reported	0	1
8794	MT-ATP6	Exercise Endurance / Coronary Atherosclerosis risk	C8794T	C-T	H-Y	+	-	Reported	1255	2
8795	MT-ATP6	MILS protective factor	A8795G	A-G	H-R	-	+	Reported	0	1
8836	MT-ATP6	LHON	A8836G	A-G	M-V	+	-	Reported	117	2
8851	MT-ATP6	BSN / Leigh syndrome	T8851C	T-C	W-R	+	+	Cfrm	3	5

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
8890	MT-ATP6	Juvenile-onset metabolic syndrome	A8890G	A-G	K-E	-	+	Reported	0	1
8932	MT-ATP6	Prostate Cancer / Neuromuscular disorder	C8932T	C-T	P-S	+	-	Reported	146	3
8950	MT-ATP6	LDYT	G8950A	G-A	V-I	+	-	Reported	56	2
8969	MT-ATP6	Mitochondrial myopathy, lactic acidosis and sideroblastic anemia (MLASA)	G8969A	G-A	S-N	-	+	Reported	0	1
8993	MT-ATP6	NARP / Leigh Disease / MILS / other	T8993C	T-C	L-P	-	+	Cfrm	2	29
8993	MT-ATP6	NARP / Leigh Disease / MILS / other	T8993G	T-G	L-R	-	+	Cfrm	6	89
9016	MT-ATP6	LHON	A9016G	A-G	I-V	-	+	Reported	6	2
9035	MT-ATP6	Ataxia syndromes	T9035C	T-C	L-P	+	+	Cfrm	0	2
9055	MT-ATP6	PD protective factor	G9055A	G-A	A-T	+	-	Reported	1887	2
9058	MT-ATP6	Possibly LVNC cardiomyopathy-associated	A9058G	A-G	T-A	+	-	Reported	18	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
9071	MT-ATP6	Potentially functional variant cosegregating with LHON3635A	C9071T	C-T	S-L	+	-	Reported	10	1
9098	MT-ATP6	Predisposition to anti-retroviral mito disease	T9098C	T-C	I-T	+	-	Reported	50	1
9101	MT-ATP6	LHON	T9101C	T-C	I-T	+	-	Reported	35	4
9127	MT-ATP6	NARP	9127-9128delAT	AT-del	IL-PTer	-	+	Reported	0	1
9134	MT-ATP6	Hypotonia, lactic acidosis, HCM, IUGR	A9134G	A-G	E-G	nr	nr	Reported	0	1
9139	MT-ATP6	LHON	G9139A	G-A	A-T	+	-	Reported - possibly synergistic	32	1
9176	MT-ATP6	FBSN / Leigh Disease	T9176C	T-C	L-P	+	+	Cfrm	3	18
9176	MT-ATP6	Leigh Disease / Spastic Paraplegia	T9176G	T-G	L-R	-	+	Cfrm	1	4

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
9185	MT-ATP6	Leigh Disease / Ataxia syndromes / NARP-like disease	T9185C	T-C	L-P	+	+	Cfrm	3	12
9191	MT-ATP6	Leigh Disease	T9191C	T-C	L-P	-	+	Reported	0	1
9205	MT-ATP6	Encephalopathy / Seizures / Lacticacidemia	9205-9206delTA	TA-del	Ter-M	+	-	Cfrm	0	7
9267	MT-CO3	MIDD	G9267C	G-C	A-P	-	+	Reported	0	1
9379	MT-CO3	MM w lactic acidosis	G9379A	G-A	W-Ter	-	+	Reported	0	1
9387	MT-CO3	Asthenozoospermia	G9387A	G-A	V-M	-	+	Reported	0	1
9438	MT-CO3	LHON	G9438A	G-A	G-S	+	-	Conflicting reports	500	13
9478	MT-CO3	Leigh Disease	T9478C	T-C	V-A	-	+	Reported	17	1
9480	MT-CO3	Myoglobinuria	9480del15	TTTTTCTTCGCAGGA-del	FFFAG-del	-	+	Reported	0	5
9537	MT-CO3	Leigh Disease	C9537insC	C-CC	Q-frameshift	+	-	Reported	0	2
9544	MT-CO3	Sporadic bilateral optic neuropathy	G9544A	G-A	G-E	.	.	Reported	0	1
9559	MT-CO3	Rhabdomyolysis	C9559del	C-del	P-frameshift-Ter	-	+	Reported	0	1
9660	MT-CO3	LHON	A9660C	A-C	M-L	+	-	Reported	0	1
9738	MT-CO3	LHON	G9738T	G-T	A-S	+	-	Reported	0	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
9789	MT-CO3	Myopathy	T9789C	T-C	S-P	-	+	Reported	0	1
9804	MT-CO3	LHON	G9804A	G-A	A-T	+	-	Reported	113	8
9861	MT-CO3	AD	T9861C	T-C	F-L	+	-	Reported	67	1
9952	MT-CO3	Mitochondrial Encephalopathy	G9952A	G-A	W-Ter	-	+	Reported	0	1
9957	MT-CO3	PEM / MELAS / NAION / HCM	T9957C	T-C	F-L	-	+	Reported	27	7
9972	MT-CO3	EXIT & APS2 - possible link	A9972C	A-C	I-L	-	+	Reported	1	1
10086	MT-ND3	Hypertensive end-stage renal disease	A10086G	A-G	N-D	+	-	Reported	396	3
10158	MT-ND3	Leigh Disease / MELAS	T10158C	T-C	S-P	+	+	Cfrm	0	16
10191	MT-ND3	Leigh Disease / Leigh-like Disease / ESOC	T10191C	T-C	S-P	-	+	Cfrm	0	20
10197	MT-ND3	Leigh Disease / Dystonia / Stroke / LDYT	G10197A	G-A	A-T	+	+	Cfrm	4	9
10237	MT-ND3	LHON	T10237C	T-C	I-T	+	-	Reported	59	1
10254	MT-ND3	Leigh Disease	G10254A	G-A	D-N	-	+	Reported	0	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
10398	MT-ND3	Invasive Breast Cancer risk factor; AD; PD; BD lithium response; Type 2 DM	A10398A	A-A	T-T	+	-	Reported; haplogroup HNTUVWXK2 marker	0	17
10398	MT-ND3	PD protective factor / longevity / altered cell pH / metabolic syndrome / breast cancer risk / ADHD	A10398G	A-G	T-A	+	-	Reported; haplogroup IJK marker	18887	28
10543	MT-ND4L	LHON	A10543G	A-G	H-R	-	+	Reported	0	1
10591	MT-ND4L	LHON	T10591G	T-G	F-C	-	+	Reported	0	1
10652	MT-ND4L	BD / MDD-associated	T10652C	T-C	syn	-	+	Reported	52	1
10663	MT-ND4L	LHON	T10663C	T-C	V-A	+	-	Cfrm	1	9
10680	MT-ND4L	LHON	G10680A	G-A	A-T	+	-	Reported - possibly synergistic	17	2
11084	MT-ND4	AD, PD; MELAS	A11084G	A-G	T-A	+	+	Reported; P.M.	175	6
11232	MT-ND4	CPEO	T11232C	T-C	L-P	-	+	Reported	0	3

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
11240	MT-ND4	Leigh Syndrome	C11240T	C-T	L-F	-	+	Reported	0	1
11253	MT-ND4	LHON; PD	T11253C	T-C	I-T	+	-	Reported	226	4
11365	MT-ND4	found in 1 HCM patient	T11365C	T-C	syn	+	-	Reported	102	1
11375	MT-ND4	found in 1 sCJD patient	A11375C	A-C	K-Q	+	-	Reported	0	1
11467	MT-ND4	Altered brain pH / sCJD patients	A11467G	A-G	syn	+	-	Reported	5378	3
11470	MT-ND4	MELAS	A11470C	A-C	K-N	-	+	Reported	0	1
11621	MT-ND4	CPEO, exercise intolerance	11621delTA	TA-del	frameshift	-	+	Reported	0	1
11696	MT-ND4	LHON / LDYT / DEAF / hypertension helper mut.	G11696A	G-A	V-I	+	+	Reported - possibly synergistic	236	10
11777	MT-ND4	Leigh Disease	C11777A	C-A	R-S	-	+	Cfrm	0	10
11778	MT-ND4	LHON / Progressive Dystonia	G11778A	G-A	R-H	+	+	Cfrm	118	267
11832	MT-ND4	EXIT / oncocytoma	G11832A	G-A	W-Ter	-	+	Reported	0	6
11874	MT-ND4	LHON	C11874A	C-A	T-N	+	-	Reported	0	1
11919	MT-ND4	Thyroid Cancer Cell Line	C11919T	C-T	S-F	+	-	Reported	0	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
11994	MT-ND4	Oligoasthenoteratozoospermia (OAT)	C11994T	C-T	T-I	+	-	Conflicting reports	0	2
12026	MT-ND4	DM	A12026G	A-G	I-V	+	-	Reported	210	2
12027	MT-ND4	SZ-associated	T12027C	T-C	I-T	.	.	Reported	2	2
12338	MT-ND5	DEAF1555 increased penetrance / LHON	T12338C	T-C	M-T	+	-	Conflicting reports	128	7
12361	MT-ND5	Non-alcoholic fatty liver disease	A12361G	A-G	T-A	+	-	Reported	239	1
12372	MT-ND5	Altered brain pH / sCJD patients	G12372A	G-A	syn	+	-	Reported	5778	3
12397	MT-ND5	PD, early onset	A12397G	A-G	T-A	+	-	Reported	276	2
12425	MT-ND5	Mitochondrial Myopathy & Renal Failure	A12425del	A-del	N-frameshift	-	+	Reported	2	1
12477	MT-ND5	possible HCM susceptibility	T12477C	T-C	syn	+	-	Reported	238	1
12622	MT-ND5	Leigh Disease	G12622A	G-A	V-I	+	+	Significance unclear	10	1
12631	MT-ND5	found in 2 sCJD patients	T12631A	T-A	S-T	+	-	Reported	0	2
12634	MT-ND5	Thyroid Cancer Cell Line	A12634G	A-G	I-V	+	-	Reported	122	2

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
12706	MT-ND5	Leigh Disease	T12706C	T-C	F-L	-	+	Cfrm	0	9
12770	MT-ND5	MELAS	A12770G	A-G	E-G	-	+	Reported	1	4
12782	MT-ND5	LHON	T12782G	T-G	I-S	-	+	Reported	0	1
12811	MT-ND5	Possible LHON factor	T12811C	T-C	Y-H	+	-	Reported	460	4
12848	MT-ND5	LHON	C12848T	C-T	A-V	-	+	Reported	0	3
13042	MT-ND5	Optic neuropathy/ retinopathy/ LD	G13042A	G-A	A-T	-	+	Cfrm	1	6
13045	MT-ND5	MELAS / LHON / Leigh overlap syndrome	A13045C	A-C	M-L	-	+	Reported	0	4
13046	MT-ND5	LHON/ MELAS overlap syndrome	T13046C	T-C	M-T	-	+	Reported	0	1
13051	MT-ND5	LHON	G13051A	G-A	G-S	+	-	Cfrm	0	2
13063	MT-ND5	Adult-onset Encephalopathy / Ataxia	G13063A	G-A	V-I	-	+	Reported	2	3
13084	MT-ND5	MELAS / Leigh Disease	A13084T	A-T	S-C	-	+	Reported	0	4
13094	MT-ND5	Ataxia+ PEO / MELAS, LD, LHON, myoclonus, fatigue	T13094C	T-C	V-A	+	+	Cfrm	1	6

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
13135	MT-ND5	possible HCM susceptibility	G13135A	G-A	A-T	+	-	Reported	400	1
13271	MT-ND5	Exercise intolerance (EXIT)	T13271C	T-C	L-P	-	+	Reported	1	2
13379	MT-ND5	LHON	A13379C	A-C	N-S	+	-	Reported	0	1
13511	MT-ND5	Leigh-like syndrome	A13511T	A-T	K-M	-	+	Reported	0	1
13513	MT-ND5	Leigh Disease / MELAS / LHON-MELAS Overlap Syndrome	G13513A	G-A	D-N	-	+	Cfrm	1	31
13514	MT-ND5	Leigh Disease / MELAS	A13514G	A-G	D-G	-	+	Cfrm	0	12
13528	MT-ND5	LHON-like, LHON, MELAS	A13528G	A-G	T-A	+	-	Reported	38	4
13580	MT-ND5	Thyroid Cancer	C13580G	C-G	A-G	-	+	Reported	0	1
13637	MT-ND5	Possible LHON factor	A13637G	A-G	Q-R	+	-	Reported	329	2
13708	MT-ND5	LHON / Increased MS risk / higher freq in PD-ADS	G13708A	G-A	A-T	+	-	P.M. - haplogroup J marker	3091	47
13730	MT-ND5	LHON	G13730A	G-A	G-E	-	+	Reported	0	7

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
13831	MT-ND5	Thyroid Cancer Cell Line	C13831A	C-A	L-M	-	+	Reported	3	1
13849	MT-ND5	MELAS	A13849C	A-C	N-H	+	-	Reported - possible secondary	0	1
13967	MT-ND5	Possible LHON factor	C13967T	C-T	T-M	+	-	Reported	116	3
14063	MT-ND5	Potentially functional variant cosegregating with LHON3635A	T14063C	T-C	I-T	+	-	Reported	24	1
14091	MT-ND5	Developmental delay, seizure, hearing loss, diabetes	A14091T	A-T	K-N	-	+	Reported	0	1
14163	MT-ND6	Possible deafness factor	C14163T	C-T	A-T	+	-	Conflicting reports	12	2
14279	MT-ND6	LHON	G14279A	G-A	S-L	+	-	Reported	6	2
14319	MT-ND6	PD, early onset	T14319C	T-C	N-D	+	-	Reported	53	2
14325	MT-ND6	LHON	T14325C	T-C	N-D	+	-	Reported	46	1
14340	MT-ND6	SNHL	C14340T	C-T	V-M	+	-	Reported	20	1
14430	MT-ND6	Thyroid Cancer	A14430G	A-G	W-R	+	-	Reported	0	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
14439	MT-ND6	Mitochondrial Respiratory Chain Disorder	G14439A	G-A	P-S	+	-	Reported	0	1
14453	MT-ND6	MELAS / Leigh Disease	G14453A	G-A	A-V	-	+	Reported	0	5
14459	MT-ND6	LDYT / Leigh Disease	G14459A	G-A	A-V	+	+	Cfrm	3	25
14482	MT-ND6	LHON	C14482A	C-A	M-I	+	+	Cfrm	2	11
14482	MT-ND6	LHON	C14482G	C-G	M-I	+	+	Cfrm	0	5
14484	MT-ND6	LHON	T14484C	T-C	M-V	+	+	Cfrm	51	149
14487	MT-ND6	Dystonia / Leigh Disease / Ataxia / Ptosis / Epilepsy	T14487C	T-C	M-V	-	+	Cfrm	0	21
14495	MT-ND6	LHON	A14495G	A-G	L-S	-	+	Cfrm	1	7
14498	MT-ND6	LHON	T14498C	T-C	Y-C	+	+	Reported	0	4
14502	MT-ND6	LHON	T14502C	T-C	I-V	+	-	Reported - possibly synergistic	154	4
14568	MT-ND6	LHON	C14568T	C-T	G-S	+	-	Cfrm	6	9
14577	MT-ND6	MIDM	T14577C	T-C	I-V	-	+	Reported	406	1
14596	MT-ND6	LHON	A14596T	A-T	I-M	+	-	Reported	0	4
14600	MT-ND6	Leigh Disease w/optic atrophy	G14600A	G-A	P-L	+	+	Reported	0	3

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
14668	MT-ND6	Depressive Disorder associated	C14668T	C-T	syn	+	-	Reported	1798	1
14787	MT-CYB	PD / MELAS	14787delTTAA	TTAA-del	I-frame shift	-	+	Reported	0	1
14831	MT-CYB	LHON	G14831A	G-A	A-T	+	-	Reported	94	1
14841	MT-CYB	LHON helper mut.	A14841G	A-G	N-S	-	+	Reported	8	1
14846	MT-CYB	EXIT	G14846A	G-A	G-S	-	+	Reported	0	5
14849	MT-CYB	EXIT / Septo-Optic Dysplasia	T14849C	T-C	S-P	-	+	Cfrm	0	3
14864	MT-CYB	MELAS	T14864C	T-C	C-R	-	+	Cfrm	2	1
15024	MT-CYB	Possible DEAF modifier	G15024A	G-A	C-Y	+	-	Reported	23	1
15043	MT-CYB	MDD-associated	G15043A	G-A	syn	+	-	Reported	9992	2
15059	MT-CYB	MM	G15059A	G-A	G-Ter	-	+	Reported	0	2
15077	MT-CYB	DEAF	G15077A	G-A	E-K	+	-	Reported	89	2
15084	MT-CYB	EXIT	G15084A	G-A	W-Ter	-	+	Reported	0	2
15092	MT-CYB	MELAS	G15092A	G-A	G-S	-	+	Reported	0	1
15150	MT-CYB	EXIT	G15150A	G-A	W-Ter	-	+	Reported	0	1
15168	MT-CYB	EXIT	G15168A	G-A	W-Ter	-	+	Reported	0	2
15170	MT-CYB	EXIT	G15170A	G-A	G-Ter	-	+	Reported	0	1
15197	MT-CYB	EXIT	T15197C	T-C	S-P	-	+	Reported	0	2
15209	MT-CYB	Prader-Willi syndrome	T15209C	T-C	Y-H	+	-	Reported	4	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
15237	MT-CYB	Potentially functional variant cosegregating with LHON3635A	T15237C	T-C	I-T	+	-	Reported	5	1
15242	MT-CYB	Mitochondrial Encephalomyopathy	G15242A	G-A	G-Ter	-	+	Reported	0	2
15243	MT-CYB	HCM	G15243A	G-A	G-E	-	+	Reported	0	3
15257	MT-CYB	LHON	G15257A	G-A	D-N	+	-	P.M. - haplogroup J2 marker - possible helper mut.	685	44
15287	MT-CYB	Possible DEAF helper mut.	T15287C	T-C	F-L	-	+	Further studies needed	70	1
15395	MT-CYB	Possible LHON factor	A15395G	A-G	K-E	+	-	Reported	2	1
15453	MT-CYB	Isolated complex III deficiency	T15453C	T-C	L-P	+	-	Reported	6	1
15497	MT-CYB	EXIT / Obesity	G15497A	G-A	G-S	+	-	Reported	187	4
15498	MT-CYB	EXIT	15498del24	24 bp deletion	GDPDNYTL-del	-	+	Reported	0	2
15498	MT-CYB	HiCM / WPW, DEAF	G15498A	G-A	G-D	-	+	Reported	12	6

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
15579	MT-CYB	Multisystem Disorder, EXIT	A15579G	A-G	Y-C	-	+	Cfrm	0	4
15615	MT-CYB	EXIT / Antimycin resistance	G15615A	G-A	G-D	-	+	Reported	0	3
15620	MT-CYB	Leigh Syndrome helper mut	C15620A	C-A	L-I	-	+	Reported	0	1
15635	MT-CYB	Polyvisceral failure	T15635C	T-C	S-P	+	-	Reported	2	1
15649	MT-CYB	Multisystem Disorder, EXIT	15649-15666del	18 bp deletion	ILAMIP-del	-	+	Reported	0	1
15662	MT-CYB	Complex mitochondriopathy-associated	A15662G	A-G	I-V	+	+	Reported	168	1
15674	MT-CYB	LHON	T15674C	T-C	S-P	+	-	Reported	140	2
15693	MT-CYB	Possibly LVNC cardiomyopathy-associated	T15693C	T-C	M-T	+	-	Reported	460	1
15699	MT-CYB	Muscle Weakness SNHL and Migraine	G15699C	G-C	R-P	-	+	Reported	0	2
15723	MT-CYB	EXIT	G15723A	G-A	W-Ter	-	+	Reported	0	1
15761	MT-CYB	MM	G15761A	G-A	G-Ter		+	Reported	0	1
15762	MT-CYB	MM	G15762A	G-A	G-E	-	+	Reported	0	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
15773	MT-CYB	LHON	G15773A	G-A	V-M	+	-	Possibly synergistic	44	1
15784	MT-CYB	POAG - potential for association	T15784C	T-C	syn	+	-	Reported	1574	3
15800	MT-CYB	EXIT / Myopathy	C15800T	C-T	Q-Ter	-	+	Reported	0	2
15812	MT-CYB	LHON	G15812A	G-A	V-M	+	-	Reported / Secondary	435	19
16081	MT-CR	Cyclic Vomiting Syndrome	A16081G	A-G	noncoding	-	+	Reported	1	1
16093	MT-CR	Cyclic Vomiting Syndrome	T16093C	T-C	noncoding	-	+	Reported	2410	1
16129	MT-CR	Cyclic Vomiting Syndrome with Migraine	G16129A	G-A	noncoding	-	+	Reported	5373	1
16176	MT-CR	Cyclic Vomiting Syndrome with Migraine	C16176T	C-T	noncoding	-	+	Reported	274	1
16183	MT-CR	Melanoma patients	A16183C	A-C	noncoding	.	.	Reported	5686	1

Position	Locus	Disease	Allele	Nucleotide Change	Amino Acid Change	Homoplasmy	Heteroplasmy	Status	GB Frequency	References
16189	MT-CR	Diabetes / Cardiomyopathy /cancer risk / mtDNA copy nbr / Metabolic Syndrome / Melanoma patients	T16189C	T-C	noncoding	+	-	Reported	10744	32
16192	MT-CR	Melanoma patients	C16192T	C-T	noncoding	.	.	Reported	1753	1
16270	MT-CR	Melanoma patients	C16270T	C-T	noncoding	.	.	Reported	2007	1
16300	MT-CR	BD-associated	A16300G	A-G	noncoding	+	-	Reported	199	2
16318	MT-CR	Non-alcoholic steatohepatitis - potential for association	A16318C	A-C	noncoding	.	.	Reported	86	1
16390	MT-CR	POAG - potential for association	G16390A	G-A	noncoding	+	-	Reported	2582	3
16519	MT-CR	Cyclic Vomiting Syndrome with Migraine /metastasis	T16519T	T-T	noncoding	+	-	Reported	0	4