Supplemental Table 1. Detail list of polymorphic variations of the whole mtDNA genome in primary brain tumor patients.

Region	Position	Alelle changes	n	Phenotype from Mitomap/ dbSNP database	Pathogenicity predictor (Mitimpact2)	dbSNP ID
D-Loop	16037	A>G	1	Frontotemporal dementia, Ovarian cancer	NA	rs1603225645
	16075	T>C	1	NA	Likely pathogenic	rs878991048
	16086	T>C	1	LHON, Leukemia, Familial deafness, Frontotemporal dementia, Sporadic Creutzfeldt-Jakob disease, tubulointerstitial kidney disease, cholestatic liver disease	Likely pathogenic	rs386420030
	16092	T>C	1	MELAS, Parkinson's disease, Familial deafness, SIDS, Leigh syndrome, Frontotemporal dementia, Ovarian cancer, Age-related macular degeneration, LHON, sporadic Creutzfeldt-Jakob disease, Cholestatic liver disease	Likely pathogenic	rs1556424740
	16093	T>C	4	MELAS, Parkinson's disease, Familial deafness, SIDS, Leigh syndrome, Frontotemporal dementia, Ovarian cancer, Age-related macular degeneration, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility, Infantile cardiomyopathy, Leukemia, Nasopharyngeal carcinoma, Multiple sclerosis	Likely pathogenic	rs2853511
	16108	C>T	2	Familial deafness, SIDS, Nasopharyngeal carcinoma	Likely pathogenic	rs386829271
	16111	C>T	1	MELAS, Parkinson's disease, Familial deafness, SIDS, Leigh syndrome, Frontotemporal dementia, LHON, sporadic Creutzfeldt-Jakob disease, Infantile cardiomyopathy, Leukemia, Nasopharyngeal carcinoma, Metabolic syndrome, Repeated pregnancy loss, Coronary heart disease	Likely pathogenic	rs35315169
	16129	G>A	8	MELAS, Parkinson's disease, Familial deafness, SIDS, Frontotemporal dementia, Ovarian cancer, LHON, Spermatozoa motility, Infantile cardiomyopathy, Leukemia, Nasopharyngeal carcinoma, Metabolic syndrome	Likely pathogenic	rs41534744

Table S1 Continued.

	16136	T>C	1	Familial deafness, LHON, Ovarian cancer, Spermatozoa motility, Infantile cardiomyopathy	NA	rs1603225675
	16140	T>C	3	MELAS, Familial deafness, Leigh syndrome, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility, Nasopharyngeal carcinoma, Coronary heart disease	NA	rs3134562
	16147	C>T	4	Frontotemporal dementia, LHON	NA	rs2854125
	16148	C>T	1	SIDS, LHON, Leukemia, Nasopharyngeal carcinoma	NA	rs201893071
	16153	G>A	1	Parkinson disease, SIDS, LHON, Frontotemporal dementia, Ovarian cancer, Spermatozoa motility, Infantile cardiomyopathy, Repeated pregnancy loss	Likely pathogenic	rs2853512
	16162	A>G	2	Familial deafness, LHON, Frontotemporal dementia, Ovarian cancer, Nasopharyngeal carcinoma	NA	rs41466049
	16169	C>T	1	Frontotemporal dementia, LHON, Leukemia	NA	rs878862655
	16172	T>C	5	MELAS, Parkinson disease, Alzheimer disease, Familial deafness, SIDS, Leigh syndrome, Frontotemporal dementia, Ovarian cancer, LHON, sporadic Creutzfeldt-Jakob disease, Cholestatic liver disease, Spermatozoa motility, Nasopharyngeal carcinoma	NA	rs2853817
	16173	C>T	1	LHON, Frontotemporal dementia, Cholestatic liver disease, Spermatozoa motility	NA	rs1556424780
	16177	A>G	1	LHON	NA	rs1556424782
	16182	A>G	1	NA	NA	rs879044186
	16192	C>T	1	MELAS, Parkinson disease, Familial deafness, SIDS, Frontotemporal dementia, Ovarian cancer, Age-related macular degeneration, LHON, sporadic Creutzfeldt-Jakob disease, Infantile cardiomyopathy, Leukemia, Nasopharyngeal carcinoma	NA	rs879025248
	16193	C>T	1	MELAS, Parkinson disease, Familial deafness, LHON Sperm motility, Frontotemporal dementia, Repeated pregnancy loss	NA	rs879091650

Table S1 Continued.

	16209	T>C	2	MELAS, LHON, Leigh syndrome, Sporadic Creutzfeldt-Jakob disease, Leukemia, Nasopharyngeal carcinoma, Cholestatic liver disease	Likely pathogenic	rs386829278
	16214	C>A	1	NA	NA	NA
	16217	T>C	4	Alzheimer disease, Familial deafness, LHON, Infantile cardiomyopathy, Leukemia, Nasopharyngeal carcinoma, Cholestatic liver disease	Likely pathogenic	rs35134837
	16223	T>C	8	Venous thromboembolism	NA	rs2853513
	16224	T>C	1	MELAS, Parkinson's disease, Cardiomyopathy, Spermatozoa motility, Dementia, LHON, Sporadic Creutzfeldt-Jakob disease, Leukemia	Likely pathogenic	rs386420031
	16234	C>T	2	MELAS, Familial deafness, SIDS, Frontotemporal dementia, Ovarian cancer, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility, Nasopharyngeal carcinoma	Likely pathogenic	rs368259300
	16235	A>G	3	Occipital stroke, Frontotemporal dementia, LHON, Leukemia	NA	rs376167878
	16243	T>C	1	Familial deafness, Leigh syndrome, Frontotemporal dementia, LHON, sporadic Creutzfeldt-Jakob disease	NA	rs386829289
	16256	C>T	3	MELAS, Parkinson’s disease, SIDS, Ovarian cancer, Age-related macular degeneration, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility, Infantile cardiomyopathy, Leukemia, Cholestatic liver disease	Likely pathogenic	rs2857289
	16257	C>A	1	MELAS, Parkinson’s disease, Familial deafness, LHON, Sporadic Creutzfeldt-Jakob disease, Cholestatic liver disease	Likely pathogenic	rs376682258
	16259	C>T	1	Frontotemporal dementia, Leukemia	NA	rs1556424830
	16261	C>T	6	MELAS, Parkinson’s disease, Familial deafness, SIDS, Leigh syndrome, Frontotemporal dementia, Ovarian cancer, Age-related macular degeneration, LHON, sporadic Creutzfeldt-Jakob disease, Cholestatic liver disease, Metabolism disease, Nasopharyngeal carcinoma	Likely pathogenic	rs138126107

Table S1 Continued.

	16263	T>C	3	Familial deafness, Age-related macular degeneration, LHON	Likely pathogenic	rs386829294
	16266	C>T	2	MELAS, Parkinson's disease, Familial deafness, Frontotemporal dementia, LHON, Nasopharyngeal carcinoma, Cholestatic liver disease	Likely pathogenic	rs879098011
	16270	C>T	1	Parkinson's disease, Familial deafness, SIDS, Frontotemporal dementia, Ovarian cancer, Age-related macular degeneration, LHON, Infantile cardiomyopathy, Leukemia, Multiple sclerosis, Malignant melanoma, Occipital stroke, Schizophrenic	NA	rs2857290
	16274	G>A	3	Prostate cancer, MELAS, Friedreich's ataxia, Deafness, Spermatozoa motility, Frontotemporal dementia, LHON, Nasopharyngeal carcinoma, Cholestatic liver disease	Likely pathogenic	rs144095641
	16278	C>T	4	MELAS, Parkinson's disease, Familial deafness, LHON, Frontotemporal dementia, Ovarian cancer, Spermatozoa motility, Infantile cardiomyopathy, Schizophrenic, Leukemia	NA	rs41458645
	16284	A>G	1	Frontotemporal dementia, LHON, sporadic mitochondrial myopathies	NA	rs1556424845
	16290	C>T	3	Breast cancer, Ovarian cancer, Parkinson's disease, LHON, Deafness, Leukemia, Nasopharyngial carcinoma, Leigh disease, Infantile cardiomyopathy, Sporadic Creutzfeldt-Jakob disease, Cholestatic liver disease	Likely pathogenic	rs1556424849
	16291	C>T	7	MELAS, Parkinson's disease, Familial deafness, SIDS, Frontotemporal dementia, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility	Likely pathogenic	rs35302802
	16293	A>C	1	Familial deafness, SIDS, LHON, Leukemia, Cholestatic liver disease	NA	rs878890610
	16294	C>G	1	NA	NA	rs140662392
	16295	C>T	3	MELAS, Leigh syndrome, LHON, Nasopharyngeal carcinoma, Occipital stroke, Anterior ischemic optic neuropathy	NA	rs878874012

Table S1 Continued.

	16297	T>C	1	Familial deafness, Frontotemporal dementia, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility, Nasopharyngeal carcinoma	NA	rs386829303
	16298	T>C	1	Stroke, Prostate cancer, Deafness, Parkinson's disease, SIDS, LHON, Leukemia, Nasopharyngeal carcinoma, Alzheimer's disease	NA	rs148377232
	16304	T>C	7	Prostate cancer, Esophageal squamous cell carcinoma, Breast cancer	Likely pathogenic	rs386829305
	16311	T>C	4	Prostate cancer, MELAS, Friedreich's ataxia, Deafness, Spermatozoa motility, Frontotemporal dementia, LHON, Nasopharyngeal carcinoma, Cholestatic liver disease	Likely pathogenic	rs34799580
	16319	G>A	5	Mitochondrial inheritance, MELAS, Parkinson's disease, Familial deafness, Frontotemporal dementia, LHON, Nasopharyngeal carcinoma, Cholestatic liver disease	NA	rs35105996
	16320	C>T	1	MELAS, Parkinson's disease, Familial deafness, SIDS, Frontotemporal dementia, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility	NA	rs62581338
	16327	C>T	1	Familial deafness, Frontotemporal dementia, LHON, sporadic Creutzfeldt-Jakob disease, Spermatozoa motility, Nasopharyngeal carcinoma	NA	rs41355449
	16343	A>G	2	LHON, Multiple sclerosis, Repeated pregnancy loss	NA	rs374065731
	16352	T>C	1	LHON, Frontotemporal dementia	NA	rs1556424871
	16362	T>C	22	Parkinson disease, Stroke, Familial deafness, SIDS, Leigh syndrome, LHON, sporadic Creutzfeldt-Jakob disease, Leukemia, Nasopharyngeal carcinoma, Multiple sclerosis, Cholestatic liver disease	NA	rs62581341
	16390	G>A	8	Open-angle glaucoma, Breast cancer, Ovarian cancer, LHON, Friedreich's ataxia, Leukemia, Nasopharyngeal carcinoma, Alzheimer's disease	NA	rs41378955

Table S1 Continued.

	16519	T>C	43	Glioblastoma, Prostate cancer, Gastric cancer, Ovarian cancer, MELAS, Occipital stroke, SIDS, Deafness, Leigh syndrome, LHON, Sporadic Creutzfeldt-Jakob disease	Likely pathogenic	rs3937033
	63	T>C	1	Cardiomyopathy, LHON	NA	rs1603218281
	66	G>T	1	Congenital cataract	NA	rs1556422371
	125	T>C	1	Nasopharyngeal carcinoma, LHON	NA	rs144402189
	127	T>C	1	Nasopharyngeal carcinoma, LHON	NA	rs1556422377
	128	C>T	1	Ovarian carcinoma, Nasopharyngeal carcinoma,LHON	NA	NA
	143	G>A	3	Deafness, Ovarian cancer, Leukemia, Age-related macular degeneration, LHON, Sporadic Creutzfeldt-Jakob disease	NA	rs375589100
	146	T>C	14	Endometriosis, Ovarian carcinoma, Prostate cancer, Alzheimer's disease	NA	rs370482130
	150	C>T	8	Cervical cancer, Longevity, Thyroid cancer, Prostate cancer, Breast cancer	NA	rs62581312
	151	C>T	2	MELAS, Parkinson's disease, Ovarian cancer, LHON	NA	rs879131641
	152	T>C	15	MELAS, Parkinson disease, Familial deafness, Esophageal squamous cell carcinoma, Leigh syndrome, Ovarian cancer, Age-related macular degeneration, LHON, Spermatozoa motility, Leukemia, Nasopharyngeal carcinoma	NA	rs117135796
	153	A>G	2	Leukemia, Nasopharyngeal carcinoma, LHON, sporadic Creutzfeldt-Jakob disease	Likely pathogenic	rs370716192
	158	T>A	1	NA	NA	NA
	185	G>A	1	Thyroid tumor, Parkinson's disease, LHON, Sporadic mitochondrial myopathies, Cardiomyopathy, Leukemia, Nasopharyngeal carcinoma, Age-related macular degeneration	NA	rs879015046
	189	A>G	1	Down syndrome, Prostate cancer, Alzheimer's disease, Gastric cancer, Ovarian cancer, Mitochondrial myopathy	NA	rs371543232

Table S1 Continued.

	195	T>C	4	Schizophrenic, Bipolar disorder, Malignant melanoma, Open-angle glaucoma, Thyroid tumor, Prostate cancer, Alzheimer's disease, Ovarian cancer	NA	rs2857291
	199	T>C	6	Ovarian carcinoma, MELAS, Parkinson's disease, Deafness, Neurodegenerative brain, Esophageal squamous cell carcinoma, LHON, Nasopharyngeal carcinoma, Sporadic Creutzfeldt-Jakob disease	NA	rs72619362
	204	T>C	1	Prostate cancer, Oral cancer, Gastric cancer, Ovarian cancer	NA	rs3135032
	207	G>A	1	Prostate cancer, Oral cancer	NA	rs369669319
	210	A>G	5	Deafness, LHON, Leukemia, Sporadic Creutzfeldt-Jakob disease	NA	rs368534078
	215	A>G	1	Esophageal squamous cell carcinoma, MELAS, Parkinson's disease, LHON, Ovarian cancer, Deafness, Sporadic Creutzfeldt-Jakob disease	NA	rs879219259
	228	G>A	1	Cataract, LHON, Cardiomyopathy, Freidreich's ataxia, Ovarian cancer, Cholestatic liver disease	NA	rs41323649
	235	A>G	1	Prostate cancer, Deafness, LHON, Esophageal squamous cell carcinoma, Leukemia, Leigh syndrome, Sporadic Creutzfeldt-Jakob disease	NA	rs3937037
	249	DEL A	7	Ovarian carcinoma, Esophageal squamous cell carcinoma, Deafness, LHON	NA	NA
	269	C>T	1	NA	NA	NA
	303-315	C7TC6	19	Ovarian cancer, Breast cancer, Thyroid cancer, Head and neck lesion, Prostate cancer, Gallbladder carcinoma, Gastric cancer, Colorectal cancer	Likely pathogenic	rs878871521
		C8TC6	23			
		C9TC6	9			
	337	DEL A	1	NA	NA	NA
	447	C>G	1	Infantile cardiomyopathy	NA	rs1603218412
	463	C>T	1	NA	NA	rs2124590746

Table S1 Continued.

	482	T>C	1	Neurodegenerative brain, Leukemia, LHON, SIDS, Age-related macular degeneration, Deafness, Cardiomyopathy	NA	rs386419941
	489	T>C	28	Colorectal cancer, Ovarian cancer, Prostate cancer	NA	rs28625645
	499	G>A	1	Pulmonary edema, Endometriosis, Thyroid tumor, Prostate cancer	NA	rs3901846
	514-523	[CA]5>[CA]4	18	Mitochondrial inheritance, SIDS, MELAS, Parkinson disease, Familial deafness, Frontotemporal dementia, LHON, Nasopharyngeal carcinoma, Cholestatic liver disease, Sporadic Creutzfeldt-Jakob disease	NA	rs78907894
	523-524	DEL AC	21	Cardiomyopathy, LHON, Cholestatic liver disease	NA	NA
	568-575	C6AC>C11	3	Thyroid tumor	NA	rs1556422469
12S rRNA	663	A>G	1	Deafness, Nasopharyngeal carcinoma, LHON, Creutzfeldt-Jakob	Benign	rs56489998
	709	G>A	8	MELAS, Parkinson's disease, LHON, Alzheimer's disease, Deafness	NA	rs2853517
	723	A>G	1	Tumors, Deafness	NA	rs386828878
	827	A>G	1	Mitochondrial non-syndromic sensorineural hearing loss, Aminoglycoside-induced deafness	Pathogenic	rs28358569
	869	C>T	2	LHON, Deafness	Likely benign	rs386828880
	930	G>C	1	Deafness	Benign	rs41352944
	1005	C>T	1	LHON	Benign	rs111033179
	1041	A>G	2	Deafness, LHON, Creutzfeldt-Jakob disease, Cardiomyopathy	NA	rs58327546
	1119	T>C	3	Mitochondrial non-syndromic sensorineural hearing loss, Aminoglycoside-induced deafness	Benign	rs397515724
	1438	A>G	44	Cystic fibrosis	Benign	rs2001030
	1598	G>A	1	Mitochondrial non-syndromic sensorineural hearing loss, Aminoglycoside-induced deafness	Benign	rs3135027
16S rRNA	1736	A>G	1	Mitochondrial inheritance	NA	rs193303006
	1780	T>C	1	Schizophrenic, Infantile cardiomyopathy	NA	rs2854127
	1809	T>C	1	NA	NA	rs879152207

Table S1 Continued.

	2070	C>T	1	NA	NA	NA
	2706	G>A	1	Improved cognitive age	NA	rs2854128
	2832	A>G	1	NA	NA	NA
	3010	G>A	3	Primary open-angle glaucoma	NA	rs3928306
	3027	T>C	7	LHON	NA	rs199838004
	3045	del A	4	NA	NA	NA
tRNA-Leu1	3290	T>C	1	Juvenile MELAS	Pathogenic	rs199474665
ND1	3316	G>C	2	Leigh syndrome	Benign	rs2853516
	3339	A>C	1	NA	NA	NA
	3394	T>C	1	LHON, Leigh syndrome	Pathogenic	rs41460449
	3505	A>G	1	Leigh syndrome	Benign	rs28358585
	3537	A>G	3	LHON, Alzheimer's disease, Atypical psychosis, Creutzfeldt-Jakob	Pathogenic	rs386828911
	3552	T>A	1	Deafness, LHON, Creutzfeldt-Jakob disease, Hypertension	Benign	rs28358587
	3606	A>G	1	LHON, Creutzfeldt-Jakob disease	NA	rs28658110
	3705	G>A	7	Ovarian carcinoma, LHON, Schizophrenic, Multiple sclerosis	NA	rs386420007
	3745	G>A	1	Leigh syndrome	Benign	rs1556422777
	3796	A>G	1	Dystonia, Leigh syndrome	Pathogenic	rs28357970
	3882	G>A	1	NA	Pathogenic	rs368898108
	3970	C>T	4	LHON	Probably damaging	rs9629042
	4071	C>T	4	Ovarian carcinoma, LHON, Metabolic syndrome, MELAS	NA	rs386828933
	4086	C>T	3	Ovarian carcinoma, LHON, Deafness, Creutzfeldt-Jakob disease	NA	rs386828934
	4131	A>G	1	LHON	NA	rs1556422817
	4170	C>T	1	Ovarian carcinoma	NA	rs1603219335
	4248	T>C	8	Thyroid tumor, LHON, Leigh disease, Creutzfeldt-Jakob disease	Possibly damaging	rs9326618
tRNA-Gln	4386	T>C	1	Juvenile MELAS	Benign	rs1569483940
ND2	4491	G>A	9	Leigh syndrome	Benign	rs201172504

Table S1 Continued.

	4511	T>C	2	NA	Benign	rs1556422867
	4562	A>G	1	Creutzfeldt-Jakob disease	NA	rs1603219514
	4697	C>T	1	NA	Benign	rs386828948
	4715	A>G	2	LHON, Deafness, Creutzfeldt-Jakob disease, Cholestatic liver disease	NA	rs28357976
	4728	A>C	1	Leigh syndrome	Likely benign	rs1556422892
	4820	G>A	1	Parkinson's disease, Deafness, Atypical psychosis, Colorectal cancer	Probably damaging	rs28357977
	4824	A>G	1	Leigh syndrome	Benign	rs1556422903
	4850	C>T	2	Adult BMI	NA	rs28413696
	4907	T>C	1	NA	NA	rs1556422922
	4959	G>A	1	Leigh syndrome	Benign	rs1603219694
	4973	T>C	1	LHON	NA	rs386828957
	5108	T>C	3	MELAS, Parkinson's disease, LHON, Creutzfeldt-Jakob disease	NA	rs386419948
	5231	G>A	1	MELAS, Parkinson's disease, LHON, Cholestatic liver disease	NA	rs371345850
	5252	G>A	1	Parkinson's disease, Infantile cardiomyopathy	Possibly damaging	rs1556422965
	5294	C>T	1	NA	NA	rs1556422969
	5417	G>A	1	MELAS, Parkinson's disease, LHON, Atypical psychosis, Creutzfeldt-Jakob disease, Cholestatic liver disease, Corticobasal degeneration	Probably damaging	rs386828968
	5442	T>C	2	Leigh syndrome	Benign	rs3020601
	5465	T>C	3	MELAS, Parkinson's disease, LHON, Cholestatic liver disease	Benign	rs3902405
	5492	T>C	1	NA	NA	rs377109345
tRNA-Trp	5553	T>C	1	Juvenile MELAS	Benign	rs878853053
nc tRNA-Trp> tRNA-Ala	5580	T>C	1	Ovarian carcinoma, Cardiomyopathy, Deafness	NA	rs1556423011
tRNA-Cys	5821	G>A	1	Juvenile MELAS	Benign	rs56133209
tRNA-Tyr	5843	A>G	1	Focal segmental glomerulosclerosis and dilated cardiomyopathy, Juvenile MELAS	Pathogenic	rs118203894

Table S1 Continued.

Nc tRNA-Tyr>COXI	5894	A>G	2	LHON	NA	rs878860965
COXI	6023	G>A	1	Mitochondrial cardiomyopathy	Probably damaging	rs1603220229
	6216	T>C	1	Atypical psychosis, Deafness, LHON	Probably damaging	rs367837524
	6221	T>C	1	LHON, Alzheimer's disease, Colorectal cancer	NA	rs370472320
	6271	A>G	1	NA	Probably damaging	NA
	6338	A>G	1	Deafness, LHON, Creutzfeldt-Jakob disease	NA	rs1603220427
	6340	C>T	2	Leigh syndrome	Benign	rs1603220429
	6392	T>C	4	Ovarian carcinoma, LHON, Atypical psychosis, Deafness	Benign	rs376513041
	6413	T>C	1	Mitochondrial cardiomyopathy	Probably damaging	rs28665937
	6455	C>T	4	MELAS, Creutzfeldt-Jakob disease, LHON, Alzheimer's disease	Probably damaging	rs28516468
	6479	A>G	1	NA	NA	NA
	6515	T>C	1	NA	NA	rs878998677
	6527	A>G	1	LHON	Probably damaging	rs878906971
	6591	C>T	1	NA	Probably damaging	rs28483589
	6620	T>C	5	Ovarian carcinoma, Cholestatic liver disease	NA	rs386828989
	6710	A>G	1	Deafness	NA	rs879242511
	6719	T>C	2	MELAS, Metabolic disease, Colorectal cancer	Likely benign	rs28358872
	6752	A>G	1	Colorectal cancer, LHON	NA	rs41332953
	6755	G>A	1	NA	NA	rs386420011
	6791	A>G	1	NA	NA	rs375385380
	6960	C>T	3	Ovarian carcinoma, LHON, Creutzfeldt-Jakob disease	Probably damaging	rs386828999
	6962	G>A	4	LHON, Deafness, Creutzfeldt-Jakob disease, Metabolic syndrome	NA	rs1970771
	7049	A>G	1	Migraine	NA	rs1603220745
	7196	C>A	1	LHON, Deafness, Creutzfeldt-Jakob disease, Cholestatic liver disease	Benign	rs28358875
	7226	G>A	1	Infantile cardiomyopathy, Deafness	NA	rs369835151
	7298	A>G	1	NA	NA	rs1603220875
COXII	7598	G>A	8	Leigh syndrome	Benign	rs386420012

Table S1 Continued.

	7684	T>C	3	LHON, Creutzfeldt-Jakob disease	NA	rs386420035
	7852	G>A	1	Hypertension	Possibly damaging	rs199751156
	7853	G>A	4	Leigh syndrome	Benign	rs386420037
	7861	T>C	1	LHON, Creutzfeldt-Jakob disease	Likely benign	rs368623956
	7885	T>C	2	NA	Possibly damaging	rs386829019
	7961	T>C	3	Infantile cardiomyopathy	Probably damaging	rs373420717
	8149	A>G	3	LHON, Deafness	NA	rs386829027
	8152	G>A	1	NA	NA	rs1603221312
	8188	A>G	2	NA	NA	rs28651339
nc COXII> tRNA-Lys	8274	delCCCTCTACC	14	NA	NA	NA
tRNA-Lys	8348	A>G	1	Juvenile MELAS	Benign	rs1556423430
ATPase8	8396	A>G	1	Leigh syndrome	Benign	rs1603221454
	8440	A>G	2	Hypertrophic cardiomyopathy, LHON, Bullous pemphigoid	Probably damaging	rs386829036
	8502	A>G	1	Leigh syndrome	Benign	rs879247004
ATPase6	8584	G>A	6	Leigh syndrome	Benign	rs3135028
	8632	T>C	1	Leigh syndrome	Benign	rs1603221654
	8701	A>G	25	Leigh syndrome, Male sub-fertility, LHON	Benign	rs2000975
	8718	A>G	1	LHON	Probably damaging	rs1556423526
	8772	T>C	1	NA	NA	rs386829049
	8794	C>T	1	Leigh syndrome	Benign	rs2298007
	8838	G>A	1	NARP syndrome	Probably damaging	rs369202065
	8843	T>C	3	Leigh syndrome	Benign	rs386829053
	8958	C>T	1	NA	Probably damaging	rs1603221942
	9053	G>A	3	Leigh syndrome	Benign	rs199646902
	9064	G>A	1	Leigh syndrome	Benign	rs386420013
	9123	G>A	2	LHON	NA	rs28358270
	9180	A>G	1	Male sub-fertility	NA	rs2298011

Table S1 Continued.

	9192	G>A	1	LHON	NA	rs386829070
COXIII	9266	G>A	1	MELAS	Likely pathogenic	rs374335946
	9509	T>C	1	NA	Probably damaging	rs375478739
	9540	T>C	17	Male sub-fertility, LHON	Probably damaging	rs2248727
	9548	G>A	1	Deafness, MELAS, LHON	NA	rs386829084
	9608	A>G	1	NA	NA	rs386829087
	9617	A>G	1	NA	Probably damaging	rs2124595636
	9728	C>A	1	NA	NA	rs1603222465
	9752	C>A	1	Leigh syndrome	Benign	rs1569484321
	9755	G>A	1	Male sub-fertility	Probably damaging	rs2856985
	9758	T>C	1	Infantile cardiomyopathy, Cholestatic liver disease	NA	rs879028351
	9824	T>C	3	MELAS, Parkinson's disease, LHON, Alzheimer's disease	NA	rs28411821
	9845	T>C	1	Deafness	NA	rs368439966
	9950	T>C	2	Male sub-fertility	NA	rs3134801
	9977	T>C	1	NA	NA	rs1603222602
tRNA-Gly	10042	A>G	1	Juvenile MELAS	Benign	rs1603222643
	10056	G>A	15	NA	NA	NA
ND3	10084	T>C	1	Leigh syndrome	Benign	rs41487950
	10238	T>C	2	LHON	Probably damaging	rs193302927
	10310	G>A	2	Male infertility, LHON	NA	rs41467651
	10365	G>A	1	Leigh syndrome	Benign	rs1603222800
	10398	A>G	20	Parkinson disease, Leigh syndrome, Male infertility, Alstrom syndrome, Male Machado-Joseph disease, Colorectal cancer, Breast cancer, Prostate cancer	Benign	rs2853826
	10400	C>T	17	Male infertility, LHON	NA	rs28358278
ND4L	10527	C>T	2	NA	Probably damaging	rs1603222877
	10609	T>C	8	Leigh syndrome	Benign	rs200487531
	10611	A>G	1	Bilateral optic neuropathy	Benign	rs386829106

Table S1 Continued.

	10754	A>G	1	Hypertension	NA	rs386419971
ND4	10834	C>T	1	Ovarian carcinoma	Probably damaging	rs386829113
	10873	T>C	2	LHON, MELAS, Parkinson's disease, Deafness, Atypical psychosis	NA	rs2857284
	10927	T>C	1	Occipital stroke, Deafness	Benign	rs878989001
	11075	T>C	1	LHON, Leigh syndrome	Probably damaging	rs370409545
	11113	T>C	1	NA	Benign	rs1603223110
	11339	T>C	1	Deafness, Leigh syndrome, Creutzfeldt-Jakob disease	Benign	rs55944643
	11464	C>A	1	NA	NA	NA
	12007	G>A	2	Psychiatric disorder	Possibly damaging	rs2853497
	12030	A>G	2	Leigh syndrome	Benign	rs1556424041
	12088	C>T	1	NA	NA	rs878866559
	12091	T>C	2	Myocardial infarction, LHON, Creutzfeldt-Jakob disease	Benign	rs28415973
tRNA-His	12190	A>G	1	Tic disorders	NA	rs1603223608
tRNA-Ser2	12236	G>A	1	Juvenile MELAS	Benign	rs28359170
	12239	C>T	2	Juvenile MELAS, Mitochondrial disease	Benign	rs376062400
tRNA-Leu2	12285	T>C	1	Juvenile MELAS	Benign	rs386419957
ND5	12354	T>C	1	LHON, Deafness, Creutzfeldt-Jakob disease, Cholestatic liver disease	NA	rs386829150
	12358	A>G	3	Leigh syndrome	Benign	rs201027657
	12362	C>T	1	Leigh syndrome	Benign	rs1603223688
	12372	G>A	3	Mitochondrial disease	Likely pathogenic	rs2853499
	12405	C>T	2	LHON, Deafness, Creutzfeldt-Jakob disease	NA	rs1556424102
	12406	G>A	5	Leigh syndrome	Benign	rs28617389
	12561	G>A	1	Leigh syndrome	Possibly damaging	rs28759201
	12705	C>T	28	LHON, MELAS, Parkinson's disease, Deafness, Colorectal cancer	Benign	rs193302956
	12771	G>A	2	MELAS, Parkinson's disease, LHON, Multiple sclerosis	Probably damaging	rs878865822
	12810	A>G	1	LHON	Probably damaging	rs28359174
	12811	T>C	2	LHON, Leigh syndrome	Pathogenic	rs199974018
	12882	C>T	5	LHON, Atypical psychosis, Deafness	Benign	rs386420001

Table S1 Continued.

	12892	T>C	1	Deafness	Possibly damaging	rs1603223959
	12957	T>C	1	LHON, Deafness, Bilateral ptosis, Creutzfeldt-Jakob disease	Benign	rs879047389
	12973	C>T	1	LHON	Probably damaging	rs2068734213
	13053	C>T	1	NA	NA	NA
	13145	G>A	3	Leigh syndrome	Benign	rs386829175
	13254	T>C	7	LHON	NA	rs372462497
	13263	A>G	1	LHON, Deafness, Metabolic syndrome, Creutzfeldt-Jakob disease	Probably damaging	rs28359175
	13395	A>G	4	LHON, Sudden cardiac death	Probably damaging	rs386829179
	13437	T>C	1	Cholestatic liver disease	NA	rs386829181
	13591	ins G	9	NA	NA	NA
	13626	C>T	3	Ovarian carcinoma, LHON	NA	rs376146541
	13651	A>G	1	Leigh syndrome	Benign	rs1569484594
	13710	A>G	1	NA	NA	rs200295632
	13759	G>A	3	Leigh syndrome	Benign	rs386420024
	13928	G>C	3	Leigh syndrome	Benign	rs28359184
	14022	A>G	1	Metabolic disease, Tubulointerstitial kidney disease	Likely benign	rs878853101
	14058	C>T	1	Thyroid cancer	NA	rs201017328
	14088	T>C	3	NA	Probably damaging	rs193302974
	14097	C>T	1	NA	NA	rs1556424382
	14110	T>C	1	Leigh syndrome	Benign	rs371451099
	14142	C>T	1	Leigh syndrome	Likely benign	rs1603224552
ND6	14152	A>G	1	LHON	Probably damaging	rs28357669
	14153	T>C	1	Leigh syndrome	Likely benign	rs1603224565
	14180	T>C	1	Leigh syndrome	Benign	rs200933339
	14182	T>C	1	LHON, Deafness, Metabolic disease, Schizophrenic	NA	rs372515139
	14209	A>G	3	NA	NA	rs375812335
	14308	T>C	1	LHON, Creutzfeldt-Jakob disease, Multiple sclerosis	NA	rs28357674
	14318	T>C	1	Leigh syndrome	Benign	rs28357675

Table S1 Continued.

	14356	C>T	1	NA	NA	rs386829209
	14371	T>C	1	NA	NA	rs386829210
	14386	T>C	1	NA	Benign	rs1603224706
	14388	A>G	1	NA	Benign	rs2068741471
	14440	A>G	1	NA	NA	rs2124598294
	14560	G>A	2	LHON	NA	rs28357676
	14569	G>A	3	MELAS, Parkinson's disease, LHON, Deafness	NA	rs386420019
	14577	T>C	4	Leigh syndrome	Benign	rs386829219
	14582	A>G	1	Leigh syndrome	Benign	rs41354845
tRNA-Glu	14687	A>G	1	Juvenile MELAS	Benign	rs200189658
	14727	T>C	2	Juvenile MELAS	Benign	rs1603224847
CytB	14766	T>C	3	Familial breast cancer, Leigh syndrome	Likely pathogenic	rs193302980
	14783	T>C	17	Familial breast cancer	Likely pathogenic	rs193302982
	14978	A>G	1	Leigh syndrome	Benign	rs199997767
	14989	C>T	1	Hypertension	NA	rs373470520
	15010	A>G	2	Ovarian carcinoma	NA	rs1603225019
	15043	G>A	19	Familial breast cancer	Likely pathogenic	rs193302985
	15097	T>C	1	Obesity	Possibly damaging	rs1556424531
	15109	T>C	1	LHON	NA	rs1603225086
	15211	C>T	2	NA	NA	rs386829237
	15217	G>A	1	LHON, MELAS, Parkinson's disease, Deafness	Likely benign	rs193302989
	15229	T>C	2	Hypertension	NA	rs1569484699
	15235	A>G	3	LHON, Obesity, Creutzfeldt-Jakob disease	Benign	rs201250154
	15236	A>G	1	Leigh syndrome	Benign	rs386829239
	15263	C>T	1	Leigh syndrome	Benign	rs200455825
	15301	G>A	20	Familial breast cancer	Likely pathogenic	rs193302991
	15317	A>G	1	Leigh syndrome, male infertility	Benign	rs2853507
	15326	A>G	36	Familial breast cancer, Leigh syndrome, mitochondrial disease	Likely pathogenic	rs2853508

Table S1 Continued.

	15346	G>A	5	Familial breast cancer	Likely pathogenic	rs527236180
	15481	C>T	1	Deafness	Benign	rs1603225305
	15487	A>T	1	Obesity, LHON, Deafness, Creutzfeldt-Jakob disease	Likely benign	rs28357370
	15511	T>C	1	Ovary neoplasm	Likely pathogenic	rs527236188
	15535	C>T	1	Deafness, Colorectal cancer, Infantile cardiomyopathy, LHON	Benign	rs28357371
	15574	C>T	1	Breast cancer	Probably damaging	rs2068747839
	15586	T>C	1	Tumors, Obesity	Probably damaging	rs878971797
	15601	T>C	1	Tumors, LHON	NA	rs1603225370
	15616	C>T	1	NA	NA	rs386829253
	15658	C>T	1	NA	Probably damaging	rs1603225412
	15663	T>C	1	Leigh syndrome	Benign	rs369851331
	15670	T>C	1	Ovary neoplasm	Likely pathogenic	rs193302997
	15746	A>G	1	Leigh syndrome	Benign	rs386829260
	15853	C>T	1	NA	Benign	rs1603225546
tRNA-Thr	15894	G>A	1	Juvenile MELAS	Benign	rs1569484752

NA: Not available; MELAS: Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes; LHON: Leber's hereditary optic neuropathy; SIDS: Sudden infant death syndrome; BMI: Body mass index; NARP: Neuropathy, ataxia, retinitis pigmentosa.