MutationsRNA lt MITOMAP lt Foswiki

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
582	MT-TF	Mitochondrial myopathy	T582C	tRNA Phe	-	+	Reported	MitoTIP72.90%	0	2
583	MT-TF	MELAS / MM & EXIT	G583A	tRNA Phe	-	+	Cfrm	MITOMAP Pathogenic	0	3
586	MT-TF	Extrapyramidal disorder with akinesia-rigidity, psychosis and SNHL	G586A	tRNA Phe	-	+	Reported	MitoTIP89.70%	0	2
602	MT-TF	Axial myopathy with encephalopathy	C602T	tRNA Phe	-	+	Reported	MitoTIP85.90%	0	2
606	MT-TF	Myoglobinuria	A606G	tRNA Phe	+	+	Unclear	MitoTIP64.90%	14	3
608	MT-TF	Tubulo-interstitial nephritis	A608G	tRNA Phe	+	-	Reported	MitoTIP65.00%	0	2
611	MT-TF	MERRF	G611A	tRNA Phe	-	+	Reported	MitoTIP51.20%	0	3
616	MT-TF	Maternally inherited epilepsy	T616C	tRNA Phe	+	+	Reported	MitoTIP83.30%	1	1
616	MT-TF	Maternally inherited epilepsy	T616G	tRNA Phe	+	+	Reported	MitoTIP95.60%	1	1
617	MT-TF	Carotid artery stenosis	G617A	tRNA Phe	-	+	Reported	MitoTIP81.70%	0	1
618	MT-TF	MM	T618C	tRNA Phe	-	+	Reported	MitoTIP65.80%	0	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
618	MT-TF	Ptosis CPEO MM & EXIT	T618G	tRNA Phe	-	+	Reported	MitoTIP77.50%	0	1
622	MT-TF	EXIT & Deafness	G622A	tRNA Phe	-	+	Reported	MitoTIP41.50%	0	2
625	MT-TF	SNHL & Epilepsy	G625A	tRNA Phe	-	+	Reported	MitoTIP81.30%	0	1
628	MT-TF	DEAF	C628T	tRNA Phe	-	+	Reported	MitoTIP34.80%	3	1
636	MT-TF	DEAF	A636G	tRNA Phe	+	-	Reported	MitoTIP1.30%	18	3
642	MT-TF	Ataxia, PEO, deafness	T642C	tRNA Phe	-	+	Reported	MitoTIP67.60%	0	1
663	MT-RNR1	Coronary Atherosclerosis risk	A663G	12S rRNA	+	-	Reported	N/A	1266	1
669	MT-RNR1	DEAF	T669C	12S rRNA	+	-	Reported	N/A	71	4
721	MT-RNR1	Possibly LVNC-associated	T721C	12S rRNA	+	-	Reported	N/A	83	1
735	MT-RNR1	DEAF	A735G	12S rRNA	.	.	Reported	N/A	52	1
745	MT-RNR1	DEAF-associated	A745G	12S rRNA	+	-	Reported	N/A	27	1
750	MT-RNR1	SZ-associated	A750A	12S rRNA	+	-	Reported	N/A	0	3
792	MT-RNR1	Increased risk of nonsyndromic deafness	C792T	12S rRNA	+	-	Reported	N/A	3	1
801	MT-RNR1	DEAF-associated	A801G	12S rRNA	+	-	Reported	N/A	6	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
827	MT-RNR1	DEAF	A827G	12SrRNA	+	-	Conflicting reports - B4b'd marker	N/A	1060	13
839	MT-RNR1	DEAF-associated	A839G	12SrRNA	+	-	Reported	N/A	5	1
850	MT-RNR1	Possibly LVNC-associated	T850C	12SrRNA	+	-	Reported	N/A	110	1
856	MT-RNR1	LHON helper / AD / DEAF-associated	A856G	12SrRNA	+	-	Reported	N/A	19	3
869	MT-RNR1	found in 1 HCM patient	C869T	12SrRNA	+	-	Reported	N/A	66	1
921	MT-RNR1	Possibly LVNC-associated	T921C	12SrRNA	+	-	Reported	N/A	330	2
960	MT-RNR1	Possibly DEAF-associated	C960del	12SrRNA	+	-	Reported	N/A	0	1
960	MT-RNR1	Possibly DEAF-associated	C960CC	12SrRNA	+	-	Reported	N/A	243	1
961	MT-RNR1	DEAF, possibly LVNC-associated	T961C	12SrRNA	+	-	Unclear	N/A	381	6
961	MT-RNR1	DEAF / AD-associated	T961delT+/-C(n)ins	12SrRNA	+	+	Unclear	N/A	0	18
961	MT-RNR1	Possibly DEAF-associated	T961G	12SrRNA	+	-	Reported	N/A	131	2
961	MT-RNR1	DEAF	T961TC	12SrRNA	+	-	Unclear	N/A	0	11
988	MT-RNR1	Possible DEAF risk factor	G988A	12SrRNA	.	.	Reported	N/A	32	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
990	MT-RNR1	DEAF	T990C	12SrRNA	+	-	Reported	N/A	29	1
1005	MT-RNR1	DEAF	T1005C	12SrRNA	+	-	Unclear	N/A	189	4
1027	MT-RNR1	DEAF-associated	A1027G	12SrRNA	+	-	Reported	N/A	12	1
1095	MT-RNR1	SNHL	T1095C	12SrRNA	+	+	Unclear	N/A	50	12
1116	MT-RNR1	DEAF	A1116G	12SrRNA	+	-	Reported	N/A	10	2
1180	MT-RNR1	Possibly DEAF-associated	T1180G	12SrRNA	+	-	Reported	N/A	0	1
1192	MT-RNR1	DEAF-associated	C1192A	12SrRNA	+	-	Reported	N/A	6	2
1192	MT-RNR1	DEAF-associated	C1192T	12SrRNA	+	-	Reported	N/A	9	1
1226	MT-RNR1	Possibly DEAF-associated	C1226G	12SrRNA	+	-	Reported	N/A	0	1
1291	MT-RNR1	DEAF	T1291C	12SrRNA	+	-	Unclear	N/A	23	3
1310	MT-RNR1	DEAF-associated	C1310T	12SrRNA	+	-	Reported	N/A	30	1
1331	MT-RNR1	DEAF-associated	A1331G	12SrRNA	+	-	Reported	N/A	6	1
1374	MT-RNR1	DEAF-associated	A1374G	12SrRNA	+	-	Reported	N/A	1	1
1391	MT-RNR1	found in 1 HCM patient	T1391C	12SrRNA	+	-	Reported	N/A	117	1
1438	MT-RNR1	SZ-associated	A1438A	12SrRNA	+	-	Reported	N/A	0	3
1452	MT-RNR1	DEAF-associated	T1452C	12SrRNA	+	-	Reported	N/A	48	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
1453	MT-RNR1	Possible DEAF risk factor	A1453G	12S rRNA	.	.	Reported	N/A	59	1
1494	MT-RNR1	DEAF	C1494T	12S rRNA	+	-	Cfrm	N/A	4	20
1517	MT-RNR1	DEAF	A1517C	12S rRNA	-	+	Reported	N/A	0	1
1537	MT-RNR1	DEAF	C1537T	12S rRNA	+	-	Reported	N/A	4	1
1555	MT-RNR1	DEAF	A1555G	12S rRNA	+	-	Cfrm	N/A	64	117
1556	MT-RNR1	found in 1 HCM patient	C1556T	12S rRNA	+	-	Reported	N/A	4	1
1606	MT-TV	AMDF	G1606A	tRNA Val	-	+	Cfrm	MITOMAP Pathogenic	0	3
1607	MT-TV	Suspected mito disease	T1607C	tRNA Val	+	+	Reported	MitoTIP18.10%	9	1
1624	MT-TV	Leigh Syndrome	C1624T	tRNA Val	+	-	Reported	MitoTIP68.70%	0	4
1630	MT-TV	MNGIE-like disease / MELAS	A1630G	tRNA Val	-	+	Cfrm	MITOMAP Pathogenic	0	2
1642	MT-TV	MELAS	G1642A	tRNA Val	-	+	Reported	MitoTIP74.30%	0	2
1643	MT-TV	Late infantile onset fatal mito disease	A1643G	tRNA Val	+	+	Reported	MitoTIP42.00%	0	1
1644	MT-TV	LS / HCM / MELAS	G1644A	tRNA Val	-	+	Cfrm	MITOMAP Pathogenic	0	4
1644	MT-TV	Adult Leigh Syndrome	G1644T	tRNA Val	-	+	Reported	MitoTIP48.40%	0	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
1659	MT-TV	Movement Disorder	T1659C	tRNA Val	-	+	Reported	MitoTIP69.60%	0	1
2352	MT-RNR2	Possibly LVNC-associated	T2352C	16S rRNA	+	-	Reported	N/A	1132	3
2361	MT-RNR2	Possibly LVNC-associated	G2361A	16S rRNA	+	-	Reported	N/A	121	1
2639	MT-RNR2	Rare mutation in a single POAG patient	C2639A	16S rRNA	+	-	Reported	N/A	1	1
2755	MT-RNR2	Possibly LVNC-associated	A2755G	16S rRNA	+	-	Reported	N/A	193	2
2835	MT-RNR2	Rett Syndrome	C2835T	16S rRNA	-	+	Reported	N/A	38	2
3010	MT-RNR2	Cyclic Vomiting Syndrome with Migraine	G3010A	16S rRNA	+	-	Reported; also common pm	N/A	6286	6
3090	MT-RNR2	Myopathy	G3090A	16S rRNA	-	+	Reported	N/A	1	1
3093	MT-RNR2	MELAS	C3093G	16S rRNA	-	+	Reported	N/A	0	2
3111	MT-RNR2	Migraine	A3111T	16S rRNA	+	-	Reported	N/A	6	1
3196	MT-RNR2	ADPD	G3196A	16S rRNA	+	+	Reported	N/A	13	3
3236	MT-TL1	Sporadic bilateral optic neuropathy	A3236G	tRNA Leu (UUR)	.	.	Reported	MitoTIP37.80%	2	2
3242	MT-TL1	MM / HCM+renal tubular dysfunction	G3242A	tRNA Leu (UUR)	+	+	Reported	MitoTIP18.50%	0	5

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
3243	MT-TL1	MELAS / LS / DMDF / MIDD / SNHL / CPEO / MM / FSGS / ASD / Cardiac+multi-organ dysfunction	A3243G	tRNA Leu(UUR)	-	+	Cfrm	MITOMAP Pathogenic	9	345
3243	MT-TL1	MM / MELAS / SNHL / CPEO	A3243T	tRNA Leu(UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	6
3244	MT-TL1	MELAS	G3244A	tRNA Leu(UUR)	-	+	Reported	MitoTIP41.60%	6	4
3249	MT-TL1	KSS	G3249A	tRNA Leu(UUR)	-	+	Reported	MitoTIP39.30%	0	3
3250	MT-TL1	MM / CPEO	T3250C	tRNA Leu(UUR)	-	+	Reported	MitoTIP33.40%	0	11
3251	MT-TL1	MM	A3251G	tRNA Leu(UUR)	-	+	Reported	MitoTIP43.50%	0	3
3252	MT-TL1	MELAS	A3252G	tRNA Leu(UUR)	-	+	Reported	MitoTIP39.40%	0	4

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
3252	MT-TL1	EXIT	A3252T	tRNA Leu (UUR)	-	+	Reported	MitoTIP39.40%	0	1
3253	MT-TL1	Maternally inherited hypertension	T3253C	tRNA Leu (UUR)	+	-	Reported	MitoTIP0.40%	6	2
3254	MT-TL1	Gestational Diabetes (GDM)	C3254A	tRNA Leu (UUR)	-	+	Reported	MitoTIP60.80%	22	1
3254	MT-TL1	MM	C3254G	tRNA Leu (UUR)	-	+	Reported	MitoTIP60.80%	0	3
3254	MT-TL1	CPEO / poss. hypertension factor	C3254T	tRNA Leu (UUR)	+	-	Reported	MitoTIP25.30%	13	5
3255	MT-TL1	MERRF / KSS overlap	G3255A	tRNA Leu (UUR)	-	+	Reported	MitoTIP75.80%	0	3
3256	MT-TL1	MELAS	C3256T	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	14
3258	MT-TL1	MELAS / Myopathy	T3258C	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	1	5
3260	MT-TL1	MMC / MELAS	A3260G	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	10

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
3264	MT-TL1	DM	T3264C	tRNA Leu (UUR)	-	+	Reported	MitoTIP47.30%	0	3
3271	MT-TL1	PEM	T3271del	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	2
3271	MT-TL1	MELAS / DM	T3271C	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	25
3273	MT-TL1	Ocular myopathy	T3273C	tRNA Leu (UUR)	-	+	Reported	MitoTIP71.20%	0	3
3274	MT-TL1	Neuropsychiatric syndrome + cataract	A3274G	tRNA Leu (UUR)	-	+	Reported	MitoTIP77.10%	0	2
3275	MT-TL1	LHON	C3275A	tRNA Leu (UUR)	+	-	Reported	MitoTIP2.20%	1	3
3277	MT-TL1	Poss. hypertension factor	G3277A	tRNA Leu (UUR)	+	-	Reported	MitoTIP2.90%	27	1
3278	MT-TL1	Poss. hypertension factor	T3278C	tRNA Leu (UUR)	+	-	Reported	MitoTIP13.10%	13	1
3280	MT-TL1	Myopathy	A3280G	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	6

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
3287	MT-TL1	Encephalomyopathy	C3287A	tRNA Leu (UUR)	-	+	Reported	MitoTIP38.30%	0	2
3288	MT-TL1	Myopathy	A3288G	tRNA Leu (UUR)	-	+	Reported	MitoTIP36.10%	0	2
3290	MT-TL1	Poss. hypertension factor	T3290C	tRNA Leu (UUR)	+	-	Reported	MitoTIP1.40%	99	2
3291	MT-TL1	MELAS / Myopathy / Deafness +Cognitive Impairment	T3291C	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	13
3302	MT-TL1	MM	A3302G	tRNA Leu (UUR)	-	+	Cfrm	MITOMAP Pathogenic	0	10
3303	MT-TL1	MMC	C3303T	tRNA Leu (UUR)	+	+	Cfrm	MITOMAP Pathogenic	0	12
4263	MT-TI	Maternally inherited essential hypertension	A4263G	tRNA Ile	+	-	Reported	MitoTIP67.80%	1	4
4267	MT-TI	MM / CPEO	A4267G	tRNA Ile	-	+	Reported	MitoTIP71.10%	0	4
4269	MT-TI	FICP	A4269G	tRNA Ile	-	+	Reported	MitoTIP82.80%	0	9
4274	MT-TI	CPEO / Motor Neuron Disease	T4274C	tRNA Ile	-	+	Reported	MitoTIP85.50%	0	5

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
4277	MT-TI	Poss. hypertension factor	T4277C	tRNA Ile	+	-	Reported	MitoTIP8.90%	14	1
4279	MT-TI	Myoclonic epilepsy	A4279G	tRNA Ile	-	+	Reported	MitoTIP54.90%	0	1
4281	MT-TI	Recurrent Myoglobinuria	A4281G	tRNA Ile	-	+	Reported	MitoTIP87.90%	1	1
4282	MT-TI	CPEO Plus	G4282A	tRNA Ile	-	+	Reported	MitoTIP82.30%	0	1
4284	MT-TI	Varied familial presentation / spastic paraparesis	G4284A	tRNA Ile	-	+	Reported	MitoTIP35.30%	2	5
4285	MT-TI	CPEO	T4285C	tRNA Ile	-	+	Reported	MitoTIP84.80%	0	5
4289	MT-TI	Retinopathy+diabetes+dysphagia+cerebral atrophy	T4289C	tRNA Ile	-	+	Reported	MitoTIP84.30%	0	1
4290	MT-TI	Progressive Encephalopathy / PEO,myopathy	T4290C	tRNA Ile	+	+	Reported	MitoTIP47.70%	0	4
4291	MT-TI	Hypomagnesemic Metabolic Syndrome	T4291C	tRNA Ile	+	-	Reported	MitoTIP31.80%	0	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
4295	MT-TI	MHCM / Maternally inherited hypertension / Maternally inherited deafness	A4295G	tRNA Ile	+	+	Reported	MitoTIP44.00%	91	10
4296	MT-TI	Leigh Syndrome	G4296A	tRNA Ile	-	+	Reported	MitoTIP46.60%	0	3
4298	MT-TI	CPEO / MS	G4298A	tRNA Ile	-	+	Cfrm	MITOMAP Pathogenic	0	9
4300	MT-TI	MICM	A4300G	tRNA Ile	+	+	Cfrm	MITOMAP Pathogenic	0	8
4302	MT-TI	CPEO	A4302G	tRNA Ile	-	+	Reported	MitoTIP42.00%	0	1
4308	MT-TI	CPEO	G4308A	tRNA Ile	-	+	Cfrm	MITOMAP Pathogenic	0	2
4309	MT-TI	CPEO	G4309A	tRNA Ile	-	+	Reported	MitoTIP64.10%	1	3
4314	MT-TI	Poss. hypertension factor	T4314C	tRNA Ile	+	-	Reported	MitoTIP1.70%	35	1
4316	MT-TI	HCM with hearing loss / poss. hypertension factor	A4316G	tRNA Ile	+	+	Reported	MitoTIP37.10%	30	2
4317	MT-TI	FICP / poss. hypertension factor	A4317G	tRNA Ile	+	-	Reported	MitoTIP2.10%	20	8
4320	MT-TI	Mitochondrial Encephalocardiomyopathy	C4320T	tRNA Ile	-	+	Reported	MitoTIP25.60%	1	4

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
4322	MT-TI	Idiopathic Dilated Cardiomopathy	C4322CC	tRNA Ile	-	+	Reported	-	3	1
4332	MT-TQ	Encephalopathy / MELAS	G4332A	tRNA Gln	-	+	Cfrm	MITOMAP Pathogenic	0	4
4336	MT-TQ	ADPD / Hearing Loss & Migraine	T4336C	tRNA Gln	+	+	Unclear	MitoTIP37.30%	367	24
4343	MT-TQ	Poss. hypertension factor	A4343G	tRNA Gln	+	-	Reported	MitoTIP5.10%	48	1
4345	MT-TQ	Poss. hypertension factor	C4345T	tRNA Gln	+	-	Reported	MitoTIP13.20%	2	1
4353	MT-TQ	Poss. hypertension factor	T4353C	tRNA Gln	+	-	Reported	MitoTIP31.60%	17	1
4363	MT-TQ	Possibly associated w DEAF + RP + dev delay / hypertension	T4363C	tRNA Gln	+	-	Reported	MitoTIP9.50%	42	3
4369	MT-TQ	Myopathy	A4369AA	tRNA Gln	-	+	Reported	-	0	2
4372	MT-TQ	Suspected mito disease	C4372T	tRNA Gln	-	+	Reported	MitoTIP71.30%	0	1
4373	MT-TQ	Possibly LVNC-associated	T4373C	tRNA Gln	+	-	Reported	MitoTIP29.10%	8	1
4381	MT-TQ	LHON	A4381G	tRNA Gln	+	-	Reported	MitoTIP15.30%	4	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
4386	MT-TQ	Heart disease / myopathy / hypertension	T4386C	tRNA Gln	+	-	Conflicting reports	MitoTIP6.90%	145	3
4387	MT-TQ	Poss. hypertension factor	C4387A	tRNA Gln	+	-	Reported	MitoTIP12.80%	0	1
4388	MT-TQ	Poss. hypertension factor	A4388G	tRNA Gln	+	-	Reported	MitoTIP0.10%	43	1
4392	MT-TQ	Poss. hypertension factor	C4392T	tRNA Gln	+	-	Reported	MitoTIP15.70%	17	1
4395	MT-TQ	Poss. hypertension factor	A4395G	tRNA Gln	+	-	Reported	MitoTIP0.20%	22	1
4401	MT-NC2	Hypertension+Ventricular Hypertrophy	A4401G	NC2 Gln-Met spacer	+	-	Reported	N/A	2	2
4403	MT-TM	Mitochondrial myopathy	G4403A	tRNA Met	-	+	Reported	MitoTIP84.80%	2	1
4409	MT-TM	Mitochondrial myopathy	T4409C	tRNA Met	-	+	Reported	MitoTIP46.50%	0	5
4410	MT-TM	Poss. hypertension factor	C4410A	tRNA Met	+	-	Reported	MitoTIP32.90%	0	1
4415	MT-TM	EXIT & APS2	A4415G	tRNA Met	-	+	Reported	MitoTIP44.10%	0	1
4435	MT-TM	LHON modulator / hypertension	A4435G	tRNA Met	+	-	Reported	MitoTIP13.80%	44	6

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
4437	MT-TM	Hypotonia, seizure, muscle weakness, lactic acidosis, hearing loss	C4437T	tRNA Met	+	-	Reported	MitoTIP67.20%	1	1
4450	MT-TM	Myopathy	G4450A	tRNA Met	-	+	Reported	MitoTIP83.70%	0	2
4454	MT-TM	Possible contributor to mito dysfunction / hypertension	T4454C	tRNA Met	+	-	Reported	MitoTIP0.80%	198	3
4456	MT-TM	Poss. hypertension factor	C4456T	tRNA Met	-	+	Reported	MitoTIP32.00%	5	1
5514	MT-TW	Neonatal onset mito disease	A5514G	tRNA Trp	+	-	Reported	MitoTIP19.70%	27	1
5521	MT-TW	Mitochondrial myopathy	G5521A	tRNA Trp	-	+	Cfrm	MITOMAP Pathogenic	0	4
5522	MT-TW	Mitochondrial myopathy	G5522A	tRNA Trp	-	+	Reported	MitoTIP83.00%	0	1
5523	MT-TW	Leigh Syndrome	T5523G	tRNA Trp	-	+	Reported	MitoTIP80.90%	0	1
5532	MT-TW	Gastrointestinal Syndrome	G5532A	tRNA Trp	-	+	Reported	MitoTIP19.40%	1	2
5537	MT-TW	Leigh Syndrome	A5537insT	tRNA Trp	-	+	Cfrm	-	0	5
5538	MT-TW	Encephalomyopathy	G5538A	tRNA Trp	-	+	Reported	MitoTIP76.70%	0	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
5540	MT-TW	Encephalomyopathy / DEAF	G5540A	tRNA Trp	-	+	Reported	MitoTIP73.70%	0	3
5541	MT-TW	MELAS+ stroke-like episodes and cortical blindness +MRI shows occipital lobe infarct	C5541T	tRNA Trp	-	+	Reported	MitoTIP84.30%	0	1
5543	MT-TW	Mitochondrial myopathy	T5543C	tRNA Trp	-	+	Reported	MitoTIP47.30%	0	5
5545	MT-TW	HCM severe multisystem disorder	C5545T	tRNA Trp	-	+	Reported	MitoTIP53.00%	0	1
5549	MT-TW	DEMCHO	G5549A	tRNA Trp	-	+	Reported	MitoTIP83.30%	0	1
5556	MT-TW	Combined OXPHOS defects	G5556A	tRNA Trp	-	+	Reported	MitoTIP44.50%	0	1
5556	MT-TW	Mito encephalomyopathy	G5556C	tRNA Trp	-	+	Reported	MitoTIP44.50%	0	1
5559	MT-TW	Leigh Syndrome	A5559G	tRNA Trp	-	+	Reported	MitoTIP70.10%	0	1
5567	MT-TW	Myopathy	T5567C	tRNA Trp	-	+	Reported	MitoTIP32.70%	36	2
5568	MT-TW	DEAF	A5568G	tRNA Trp	+	-	Reported	MitoTIP9.70%	6	1
5587	MT-TA	Possible DEAF modifier	T5587C	tRNA Ala	+	-	Reported	MitoTIP12.10%	25	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
5591	MT-TA	Myopathy	G5591A	tRNA Ala	-	+	Reported	MitoTIP68.40%	0	2
5592	MT-TA	Coronary Heart Disease	A5592G	tRNA Ala	+	-	Reported	MitoTIP0.40%	23	2
5613	MT-TA	CPEO	T5613C	tRNA Ala	-	+	Reported	MitoTIP59.30%	0	1
5628	MT-TA	CPEO / DEAF enhancer	T5628C	tRNA Ala	-	+	Reported	MitoTIP78.90%	78	3
5636	MT-TA	PEO	T5636C	tRNA Ala	-	+	Reported	MitoTIP73.50%	0	1
5650	MT-TA	Myopathy	G5650A	tRNA Ala	-	+	Cfrm	MITOMAP Pathogenic	1	4
5655	MT-TA	DEAF enhancer	T5655C	tRNA Ala	+	-	Reported	MitoTIP26.70%	292	1
5658	MT-TN	Mitochondrial myopathy	T5658C	tRNA Asn	-	+	Reported	MitoTIP94.30%	0	1
5690	MT-TN	CPEO+ptosis+proximal myopathy	A5690G	tRNA Asn	-	+	Cfrm	MITOMAP Pathogenic	0	2
5692	MT-TN	CPEO / MM	T5692C	tRNA Asn	-	+	Reported	MitoTIP46.60%	0	4
5693	MT-TN	Encephalomyopathy	T5693C	tRNA Asn	+	-	Reported	MitoTIP31.20%	0	1
5698	MT-TN	CPEO / MM	G5698A	tRNA Asn	-	+	Reported	MitoTIP47.70%	1	4
5703	MT-TN	CPEO / MM	G5703A	tRNA Asn	-	+	Cfrm	MITOMAP Pathogenic	0	5
5709	MT-TN	Ophthalmoparesis+respiratory impairment	T5709C	tRNA Asn	-	+	Reported	MitoTIP49.80%	0	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
5728	MT-TN	Multiorgan failure	T5728C	tRNA Asn	-	+	Reported	MitoTIP70.80%	1	1
5780	MT-TC	SNHL	G5780A	tRNA Cys	-	+	Reported	MitoTIP35.50%	11	1
5783	MT-TC	Myopathy deafness	G5783A	tRNA Cys	-	+	Reported	MitoTIP66.90%	30	1
5802	MT-TC	DEAF1555 increased penetrance	T5802C	tRNA Cys	+	-	Reported	MitoTIP58.90%	0	2
5814	MT-TC	Mitochondrial Encephalopathy	T5814C	tRNA Cys	-	+	L2b marker	MitoTIP38.80%	126	9
5816	MT-TC	Progressive Dystonia	A5816G	tRNA Cys	+	-	Reported	MitoTIP59.90%	0	3
5821	MT-TC	DEAF helper mut.	G5821A	tRNA Cys	+	-	Reported	MitoTIP20.90%	246	4
5843	MT-TY	FSGS / Mitochondrial Cytopathy	A5843G	tRNA Tyr	+	-	Reported	MitoTIP8.40%	196	1
5874	MT-TY	EXIT	T5874G	tRNA Tyr	-	+	Reported	MitoTIP38.90%	0	1
7445	MT-TS1 precursor	DEAF	A7445C	tRNA Ser (UCN) precursor	+	-	Reported	-	10	4
7445	MT-TS1 precursor	SNHL	A7445G	tRNA Ser (UCN) precursor	+	+	Cfrm	-	1	29

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
7445	MT-TS1 precursor	SNHL	A7445T	tRNA Ser (UCN) precursor	+	-	Reported	-	2	1
7451	MT-TS1	CPEO+ptosis	A7451T	tRNA Ser (UCN) precursor	-	+	Reported	MitoTIP80.70%	0	1
7453	MT-TS1	Fatal neonatal lactic acidosis	G7453A	tRNA Ser (UCN)	+	-	Reported	MitoTIP68.00%	0	2
7456	MT-TS1	DEAF	A7456G	tRNA Ser (UCN)	+	-	Unclear	MitoTIP16.00%	1	1
7458	MT-TS1	PEO	G7458A	tRNA Ser (UCN)	-	+	Reported	MitoTIP86.00%	0	1
7462	MT-TS1	DEAF	C7462T	tRNA Ser (UCN)	+	-	Reported	MitoTIP11.20%	6	1
7471	MT-TS1	PEM / AMDF / Motor neuron disease-like	C7471insC	tRNA Ser (UCN)	+	+	Cfrm	-	7	27
7472	MT-TS1	MM / DMDF modulator	A7472C	tRNA Ser (UCN)	+	-	Reported	MitoTIP3.20%	2	3

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
7472	MT-TS1	PEM / AMDF / Motor neuron disease-like	A7472insC	tRNA Ser (UCN)	+	+	See 7471insC	-	0	1
7480	MT-TS1	MM	T7480G	tRNA Ser (UCN)	-	+	Reported	MitoTIP46.60%	0	3
7492	MT-TS1	Hypertension	C7492T	tRNA Ser (UCN)	+	-	Reported	MitoTIP0.10%	8	1
7497	MT-TS1	MM / EXIT	G7497A	tRNA Ser (UCN)	+	+	Cfrm	MITOMAP Pathogenic	1	6
7501	MT-TS1	Cardiovascular disease; renal disease patient	T7501A	tRNA Ser (UCN)	.	.	Reported	MitoTIP1.90%	1	3
7505	MT-TS1	Maternally inherited hearing loss	T7505C	tRNA Ser (UCN)	+	-	Reported	MitoTIP58.60%	0	1
7506	MT-TS1	PEO with hearing loss	G7506A	tRNA Ser (UCN)	-	+	Reported	MitoTIP81.40%	0	1
7510	MT-TS1	SNHL	T7510C	tRNA Ser (UCN)	-	+	Cfrm	MITOMAP Pathogenic	1	13
7511	MT-TS1	SNHL	T7511C	tRNA Ser (UCN)	+	+	Cfrm	MITOMAP Pathogenic	1	16

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
7512	MT-TS1	PEM / MERME	T7512C	tRNA Ser (UCN)	+	+	Reported	MitoTIP64.20%	0	10
7520	MT-TD	Sporadic bilateral optic neuropathy	G7520A	tRNA Asp	.	.	Reported	MitoTIP54.90%	0	1
7526	MT-TD	Mitochondrial myopathy	A7526G	tRNA Asp	-	+	Reported	MitoTIP50.40%	0	1
7539	MT-TD	Multisystemic mitochondrial disorder	C7539T	tRNA Asp	-	+	Reported	MitoTIP93.70%	0	1
7543	MT-TD	MEPR	A7543G	tRNA Asp	-	+	Reported	MitoTIP67.30%	37	1
7554	MT-TD	Myopathy+ataxia+nystagmus+migraines+lactic acidosis	G7554A	tRNA Asp	-	+	Reported	MitoTIP71.20%	1	1
8296	MT-TK	DMDF / MERRF / HCM / epilepsy	A8296G	tRNA Lys	+	+	Reported	MitoTIP72.30%	33	16
8299	MT-TK	PEO + respiratory impairment	G8299A	tRNA Lys	-	+	Reported	MitoTIP63.80%	0	1
8302	MT-TK	Encephalopathy	A8302T	tRNA Lys	+	-	Unclear	MitoTIP15.20%	0	1
8304	MT-TK	Epilepsy+ataxia+visual disturbance+deafness	G8304A	tRNA Lys	-	+	Reported	MitoTIP89.70%	0	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
8306	MT-TK	Severe adult-onset multisymptom myopathy	T8306C	tRNA Lys	-	+	Reported	MitoTIP88.80%	0	2
8311	MT-TK	Poss. hypertension factor	T8311C	tRNA Lys	+	-	Reported	MitoTIP6.80%	46	1
8313	MT-TK	MNGIE / Progressive mito cytopathy	G8313A	tRNA Lys	-	+	Reported	MitoTIP73.60%	1	5
8316	MT-TK	MELAS	T8316C	tRNA Lys	-	+	Reported	MitoTIP80.20%	0	3
8319	MT-TK	Kearns-Sayre syndrome	A8319G	tRNA Lys	-	+	Reported	MitoTIP69.60%	0	1
8326	MT-TK	Mitochondrial Cytopathy	A8326G	tRNA Lys	-	+	Reported	MitoTIP46.20%	0	3
8328	MT-TK	Mito Encephalopathy / EXIT with myopathy and ptosis	G8328A	tRNA Lys	-	+	Reported	MitoTIP83.30%	0	5
8332	MT-TK	Dystonia and stroke-like episodes	A8332G	tRNA Lys	+	-	Reported	MitoTIP62.80%	0	1
8337	MT-TK	Poss. hypertension factor	T8337C	tRNA Lys	+	-	Reported	MitoTIP6.80%	167	1
8340	MT-TK	Myopathy / Exercise Intolerance / Eye disease+ SNHL	G8340A	tRNA Lys	-	+	Reported	MitoTIP64.60%	0	5

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
8342	MT-TK	PEO and Myoclonus	G8342A	tRNA Lys	-	+	Reported	MitoTIP77.20%	0	4
8343	MT-TK	Possible PD risk factor	A8343G	tRNA Lys	+	-	Reported	MitoTIP4.70%	32	1
8344	MT-TK	MERRF; Other - LD / Depressive mood disorder / leukoencephalopathy / HiCM	A8344G	tRNA Lys	-	+	Cfrm	MITOMAP Pathogenic	3	111
8347	MT-TK	Poss. hypertension factor	A8347G	tRNA Lys	+	-	Reported	MitoTIP2.60%	18	2
8348	MT-TK	Cardiomyopathy / SNHL / poss. hypertension factor	A8348G	tRNA Lys	+	+	Reported	MitoTIP33.80%	90	8
8355	MT-TK	Myopathy	T8355C	tRNA Lys	-	+	Reported	MitoTIP67.20%	0	2
8356	MT-TK	MERRF	T8356C	tRNA Lys	-	+	Cfrm	MITOMAP Pathogenic	0	10
8361	MT-TK	MERRF	G8361A	tRNA Lys	-	+	Reported	MitoTIP64.80%	0	3
8362	MT-TK	Myopathy	T8362G	tRNA Lys	-	+	Reported	MitoTIP93.00%	0	4
8363	MT-TK	MICM+DEAF / MERRF / Autism / LS / Ataxia+Lipomas	G8363A	tRNA Lys	-	+	Cfrm	MITOMAP Pathogenic	0	18

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
9997	MT-TG	MHCM	T9997C	tRNA Gly	-	+	Reported	MitoTIP80.30%	1	4
10006	MT-TG	CIPO / Encephalopathy	A10006G	tRNA Gly	+	-	Unclear	MitoTIP19.30%	6	4
10010	MT-TG	PEM	T10010C	tRNA Gly	-	+	Cfrm	MITOMAP Pathogenic	0	9
10014	MT-TG	Myopathy	G10014A	tRNA Gly	+	-	Unclear	MitoTIP60.90%	0	1
10044	MT-TG	SIDS	A10044G	tRNA Gly	-	+	Unclear	MitoTIP34.70%	116	7
10406	MT-TR	Mitochondrial myopathy	G10406A	tRNA Arg	-	+	Reported	MitoTIP72.30%	0	2
10437	MT-TR	Mitochondrial myopathy	G10437A	tRNA Arg	-	+	Reported	MitoTIP51.70%	0	1
10438	MT-TR	Progressive Encephalopathy	A10438G	tRNA Arg	-	+	Reported	MitoTIP46.20%	0	1
10450	MT-TR	Combined OXPHOS defects & severe multisystem disorder	A10450G	tRNA Arg	-	+	Reported	MitoTIP69.60%	0	1
10454	MT-TR	DEAF helper mut.	T10454C	tRNA Arg	+	-	Reported	MitoTIP4.80%	157	3
12146	MT-TH	MELAS	A12146G	tRNA His	+	+	Reported	MitoTIP61.60%	0	1
12147	MT-TH	MERRF-MELAS / Encephalopathy	G12147A	tRNA His	-	+	Cfrm	MITOMAP Pathogenic	0	5

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
12148	MT-TH	Developmental delay, optic atrophy, cataract, hearing loss, myopathy	T12148C	tRNA His	-	+	Reported	MitoTIP74.70%	1	1
12183	MT-TH	RP + DEAF	G12183A	tRNA His	-	+	Reported	MitoTIP70.30%	1	2
12187	MT-TH	Asthenozoospermia	C12187A	tRNA His	+	-	Reported	MitoTIP15.40%	0	1
12192	MT-TH	MICM	G12192A	tRNA His	+	-	Reported	MitoTIP4.50%	99	2
12201	MT-TH	Maternally inherited non-syndromic deafness	T12201C	tRNA His	-	+	Reported	MitoTIP66.70%	0	1
12206	MT-TH	MELAS-like encephalopathy+bilateral optic atrophy	C12206T	tRNA His	-	+	Reported	MitoTIP44.20%	0	1
12207	MT-TS2	Myopathy / Encephalopathy	G12207A	tRNA Ser (AGY)	-	+	Reported	MitoTIP76.40%	0	2
12224	MT-TS2	DEAF helper mut.	C12224T	tRNA Ser (AGY)	+	-	Reported	MitoTIP30.40%	0	1
12236	MT-TS2	DEAF	G12236A	tRNA Ser (AGY)	+	-	Reported	MitoTIP2.20%	327	4

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
12246	MT-TS2	CIPO	C12246A	tRNA Ser (AGY)	nd	nd	Reported	MitoTIP3.20%	3	2
12258	MT-TS2	DMDF / RP+SNHL	C12258A	tRNA Ser (AGY)	-	+	Cfrm	MITOMAP Pathogenic	1	6
12261	MT-TS2	Myopathy+epilepsy+retinal degeneration+DEAF	T12261C	tRNA Ser (AGY)	-	+	Reported	MitoTIP65.30%	0	1
12262	MT-TS2	Progressive MM+Deafness+Seizures	C12262A	tRNA Ser (AGY)	-	+	Reported	MitoTIP84.50%	0	1
12264	MT-TS2	Multisystem Disease with Cataracts / Myopathy+epilepsy+DEAF+atypical autism	C12264T	tRNA Ser (AGY)	+	+	Reported	MitoTIP79.30%	0	2
12276	MT-TL2	CPEO	G12276A	tRNA Leu (CUN)	-	+	Cfrm	MITOMAP Pathogenic	1	3
12283	MT-TL2	CPEO	G12283A	tRNA Leu (CUN)	-	+	Reported	MitoTIP43.20%	1	2
12294	MT-TL2	CPEO	G12294A	tRNA Leu (CUN)	-	+	Reported	MitoTIP71.40%	0	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
12297	MT-TL2	Dilated Cardiomyopathy / LS / Failure to Thrive & LA	T12297C	tRNA Leu (CUN)	+	+	Reported	MitoTIP47.30%	32	5
12299	MT-TL2	MELAS	A12299C	tRNA Leu (CUN)	-	+	Reported	MitoTIP53.00%	0	1
12300	MT-TL2	3243 suppressor mutant	G12300A	tRNA Leu (CUN)	-	+	Reported	MitoTIP51.70%	0	4
12308	MT-TL2	CPEO / Stroke / CM / Breast & Renal & Prostate Cancer Risk / Altered brain pH /sCJD	A12308G	tRNA Leu (CUN)	+	+	Hg K & U marker	MitoTIP42.00%	5359	19
12311	MT-TL2	CPEO	T12311C	tRNA Leu (CUN)	+	+	Reported	MitoTIP34.40%	52	3
12313	MT-TL2	FSHD	T12313C	tRNA Leu (CUN)	-	+	Reported	MitoTIP73.20%	0	1
12315	MT-TL2	CPEO / KSS	G12315A	tRNA Leu (CUN)	-	+	Cfrm	MITOMAP Pathogenic	0	7
12316	MT-TL2	CPEO	G12316A	tRNA Leu (CUN)	-	+	Cfrm	MITOMAP Pathogenic	0	2

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
12317	MT-TL2	CPEO+ptosis+myopathy+exercise intolerance+diabetes	T12317C	tRNA Leu (CUN)	-	+	Reported	MitoTIP41.30%	1	1
12320	MT-TL2	MM	A12320G	tRNA Leu (CUN)	-	+	Reported	MitoTIP37.30%	0	6
14674	MT-TE	Reversible COX deficiency myopathy	T14674C	tRNA Glu	+	-	Cfrm	MITOMAP Pathogenic	7	5
14674	MT-TE	Reversible COX deficiency myopathy	T14674G	tRNA Glu	+	-	Reported	MitoTIP29.40%	0	1
14680	MT-TE	Mitochondrial encephalomyopathy	C14680A	tRNA Glu	-	+	Reported	MitoTIP35.50%	0	1
14685	MT-TE	Cataracts w spastic paraparesis & ataxia	G14685A	tRNA Glu	-	+	Reported	MitoTIP77.40%	0	1
14687	MT-TE	Mito myopathy w respiratory failure	A14687G	tRNA Glu	+	-	Reported	MitoTIP7.00%	255	2
14692	MT-TE	LHON helper mut.	A14692G	tRNA Glu	+	-	Reported	MitoTIP2.40%	18	1
14693	MT-TE	MELAS / LHON / DEAF / hypertension helper	A14693G	tRNA Glu	+	+	Reported	MitoTIP39.50%	229	11

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
14696	MT-TE	Progressive Encephalopathy	A14696G	tRNA Glu	-	+	Reported	MitoTIP22.00%	39	1
14709	MT-TE	MM+DMDF / Encephalomyopathy / Dementia+diabetes+ophthalmoplegia	T14709C	tRNA Glu	+	+	Cfrm	MITOMAP Pathogenic	1	20
14710	MT-TE	Encephalomyopathy + Retinopathy	G14710A	tRNA Glu	-	+	Cfrm	MITOMAP Pathogenic	0	5
14721	MT-TE	Isolated complex I deficiency	G14721A	tRNA Glu	-	+	Reported	MitoTIP82.90%	0	1
14723	MT-TE	CPEO + Myopathy	T14723C	tRNA Glu	-	+	Reported	MitoTIP73.50%	0	2
14724	MT-TE	Mito Leukoencephalopathy	G14724A	tRNA Glu	-	+	Reported	MitoTIP88.80%	0	3
14728	MT-TE	Late-onset mitochondrial encephalomyopathy	T14728C	tRNA Glu	-	+	Reported	MitoTIP48.50%	0	1
14739	MT-TE	EXIT	G14739A	tRNA Glu	-	+	Reported	MitoTIP62.10%	0	2
15908	MT-TT	DEAF helper mut.	T15908C	tRNA Thr	+	-	Reported	MitoTIP28.00%	101	2
15915	MT-TT	Encephalomyopathy	G15915A	tRNA Thr	-	+	Reported	MitoTIP73.70%	1	2

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
15923	MT-TT	LIMM / MERRF / mito disease	A15923G	tRNA Thr	-	+	Reported	MitoTIP46.60%	0	4
15924	MT-TT	LIMM	A15924G	tRNA Thr	.	.	P.M.	MitoTIP22.70%	1516	6
15927	MT-TT	Multiple Sclerosis / DEAF1555 increased penetrance / CHD	G15927A	tRNA Thr	+	-	P.M. / possible helper mutation	MitoTIP16.20%	386	10
15928	MT-TT	Multiple Sclerosis / idiopathic repeat miscarriage / AD protection	G15928A	tRNA Thr	+	-	P.M. / possible helper mutation	MitoTIP20.20%	2047	7
15933	MT-TT	Suspected mito disease	G15933A	tRNA Thr	+	-	Reported	MitoTIP66.80%	0	1
15942	MT-TT	Possibly LVNC-associated	T15942C	tRNA Thr	+	-	Reported	MitoTIP28.60%	365	1
15944	MT-TT	MM	T15944del	tRNA Thr	+	-	P.M.	MitoTIP19.90%	614	2
15950	MT-TT	Dopaminergic nerve cell death (PD)	G15950A	tRNA Thr	+	-	Reported	MitoTIP54.50%	1	1
15951	MT-TT	LHON / LHON modulator	A15951G	tRNA Thr	+	-	Conflicting reports	MitoTIP23.70%	321	5
15965	MT-TP	Dopaminergic nerve cell death (PD)	A15965G	tRNA Pro	+	-	Reported	MitoTIP2.10%	8	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
15967	MT-TP	MERRF-like disease	G15967A	tRNA Pro	-	+	Reported	MitoTIP78.90%	0	2
15975	MT-TP	Ataxia+RP+deafness	C15975T	tRNA Pro	-	+	Reported	MitoTIP78.30%	0	1
15990	MT-TP	MM	C15990T	tRNA Pro	-	+	Reported	MitoTIP51.70%	0	4
15995	MT-TP	Mitochondrial cytopathy	G15995A	tRNA Pro	-	+	Reported	MitoTIP80.00%	0	2
15998	MT-TP	Mitochondrial myopathy	A15998T	tRNA Pro	-	+	Reported	MitoTIP57.50%	0	1
16002	MT-TP	Mitochondrial cytopathy	T16002C	tRNA Pro	-	+	Reported	MitoTIP75.80%	0	1
16015	MT-TP	Mitochondrial myopathy	T16015C	tRNA Pro	-	+	Reported	MitoTIP50.40%	0	1
16018	MT-TP	Dilated cardiomyopathy (15 bp dup), alternate notation	T16018TTCTCTGTTCTTTCAT	tRNA Pro	-	+	Reported	-	0	1
16021	MT-TP	Mitochondrial myopathy	16021_16022delCT	tRNA Pro	-	+	Reported	-	0	1
16023	MT-TP	Migraine+pigmentary retinopathy+deafness+leukariosis	G16023A	tRNA Pro	-	+	Reported	MitoTIP83.70%	0	1
16032	MT-TP	Dilated cardiomyopathy (15 bp dup)	T16032TTCTCTGTTCTTTCAT	tRNA Pro	-	+	Reported	-	1	1

Position	Locus	Disease	Allele	RNA	Homoplasmy	Heteroplasmy	Status	MitoTIP†	GB Frequency	References
16033	MT-TP	Dilated cardiomyopathy (15 bp dup), alternate notation	G16033TCTCTTGTTCTTTCATG	tRNA Pro	-	+	Reported	-	0	1