Table S1 mtDNA Mutations Reported in Common Mitochondrial Diseases

Mitochondria Disease	**Gene**	**mtDNA Mutation**	**Cited article**
LS	MT-ATP6, complex 5	m.8993T>C	[1–3]
		m.8993T>G	[1,3,4]
		m.8993A>G	[1,5]
		m.9176T>C	[6–9]
	MT-ND I, complex I	m.3460G>A	[10]
		m.3697G>A	[11]
		m.3946G>A	[12]
	MT-ND2, complex I	m.4681T>C	[10]
	I	m.4833A>G	[13]
	MT-ND3, complex I	m.10134C>A	[14]
		m.10158T>C	[12,15,16]
		m.10191T>C	[12,15,17–19]
		m.10197G>A	[20–22]
		m.10254G>A	[23]
	MT-ND4, complex I	m.11240C>T	[24]
		m.11777A>C	[19]
		m.11778G>A	[19]
	MT-ND5, complex I	m.12706T>C	[15,25]
		m.13513G>A	[12,13,19,26–29]
		m.13514T>C	[15]
		m.12338T>C	[13]
	MT-ND6, complex I	m.14439G>A	[12]
		m.14502T>C	[13]
		m.14459G>A	[12,30]
		m.14487T>C	[15,31]
	MT-COXII, complex I	m.8108A>G	[13]
	MT-COXIII, complex I	m.9537 C_{Ins}	[32]
	MT-TL I	m.3243A>G	[19]
LHON	MT-ND I, complex I	m.3376G>A	[33]
		m.3394T>C	[34]
		m.3395A>G	[35]
		m.3460G>A	[36,37]
		m.3472T>C	[38]
		m.3634A>G	[39]
		m.3635G>A	[40,41]
		m.3697G>A	[42]
		m.3700G>A	[43]
		m.3733G>A	[44]
		m.3866T>C	[45]
		m.4171C>A	[46]
	MT-ND 2, complex I	m.4640C>A	[47]
	MT-CO3, complex 4	m.9804G>A	[48,49]
	MT-ND 3, complex I	m.10197G>A	[21]
	MT-ND 4, complex I	m.10663T>C	[50]
		m.11778G>A	[51,52]

	MT-ND 5, complex I	m.13051G>A	[53]
		m.13094T>C	[54]
		m.12848T>C	[55]
	MT-ND6, complex I	m.14279G>A	[56]
		m.14459G>A	[57–59]
		m.14482C>A	[60]
		m.14482C>G	[61]
		m.14484A>G	[62–64]
		m.14495A>G	[65]
		m.14502T>C	[66]
		m.14568C>T	[67]
	tRNA	m.15927G>A	[68]
MERRF	MT-TF	m.611G>A	[69]
	MT-TL1	m.3255G>A	[70]
		m.3291T>C	[71]
	MT-TI	m.4279A>G	[72]
		m.4284G>A	[73]
	MT-TW	m.5521G>A	[74]
	MT-TS1	m.7512T>C	[75]
	MT-TK	m.8296A>G	[76]
		m. 8344A>G	[20,57,77–80]
		m.8356T>C	[81,82]
		m.8361G>A	[83]
		m.8363G>A	[81,82]
	MT-TH	m.12147G>A	[84,85]
	MT-TL2	m.12300G>A	[86]
	MT-ND 5, Complex I	m.13042A>T	[82,87]
	MT-ND6, Complex I	m.14709T>C	[88]
	MT-TP	m.15967G>A	[89]
MELAS	MT-TF	m.583G>A	[90–92]
	MT-TV	m.1616A>G	[93]
		m.1630A>G	[94]
		m.1642G>A	[95,96]
		m.1644G>A	[94,97,98]
	MT-RNR2	m.3093C>G	[99,100]
	MT-TLI	**m.3243A>G**	[57,77,78,81,101–105]
		m.3244G>A	[106–108]
		m.3252A>G	[92,109]
		m.3256C>T	[104,105,110,111]
		m.3258T>C	[112,113]
		m.3260A>G	[114–117]
		m.3271T>C	[57,79,103–105,118]
		m.3291T>C	[92,119–121]
	MT-ND1, Complex I	m.3308T>C	[122]

		m.3376G>A	[33,123]
		m.3380G>A	[124]
		m.3481G>A	[125,126]
		m.3697G>A	[42,127]
		m.3946G>A	[127–129]
		m.3949T>C	[127,128]
		m.3959G>A	[130]
		m.3995A>G	[130]
	MT-T1	m.4290T>C	[131]
		m.4320C>T	[132]
	MT-TQ	m.4332G>A	[133]
	MT-TW	m.5540G>A	[134]
	MT-TN	m.5693T>C	[135]
	MT-TC	m.5814T>C	[136]74
		m.5816A>G	[137,138]
	MT-CO1, Complex 4	m.6597C>A	[139]
	MT-TK	m.8316T>C	[140,141]
	MT-CO3, Complex 4	m.9957T>C	[95,142]
	MT-ND 3, Complex I	m.10197G>A	[20]
	MT-TH	m.12146A>G	[143]
		m.12206C>T	[144]
	MT-TL2	m.12299A>C	[145]
	MT-ND5, Complex I	m.12770A>G	[27,146–148]
		m.13042A>T	[87,104]
		m.13045A>C	[27,146,147]
		m.13046T>C	[149]
		m.13084A>T	[27,150]
		m.13513G>A	[27,95,151]
		m.13514A>G	[152]
		m.13528A>G	[150,153]
		m.13849A>C	[154]
	MT-TE	m.14693A>G	[155]
	MT-ND6,Complex 1	m.14453G>A	[156]
	MT-CYB, Complex 3	m.14787delTTAA	[157]
		m.14864T>C	[158]
		m.15092G>A	[159]
		m.15533A>G	[160]
Pearson Syndrome	424 bp deletion from 8578 -14001		[161]
	5182 bp deletion 10,901–16,082		[162]
	4977 bp deletion from 8482–13460		[163]
	2461 bp deletion from 10368 12828		[164]
KSS	**4.9 kb deletion from 8469 to 13447**		[165]
	MT-TLI	m.3243A>G	[166]
CPEO	MT-TM	m.4414T>C	[167]
	MT-TN	m.5667G>A	[168]
	MT-TL2	m.12315G>A	[169]

NARP	MT-ATP6,complex V	m.9127–9128 del	[170]
		m.8993T>G/C	[171]
MIDD	tRNA	**m.3243A > G**	[172,173]
	MT-ND1, Complex I	m.3421A > G	[174]
	MT-CO2	m.8241 T > G	[175]

Note: Mutations indicated in bold are the primary mutations in the respective diseases

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