Supplementary data:

Compound mitochondrial DNA mutations in a neurological patient with ataxia, myoclonus and deafness

Ji Hoon Park, Bo Ram Yoon, Hye Jin Kim, Phil Hyu Lee, Byung-Ok Choi and Ki Wha Chung

J. Genet. **93**, 173–177

Table 1. Variations from the whole mtDNA sequence in the AMDF patient.

Locus/gene	Mutation		Report		Polymorphism/phenotype[d]
	Nucleotide[a]	Amino acid[b]	Mitomap/mtDB[c]	dbSNP135	
D-Loop	73A>G	–	Y/Y	rs3087742	Pol
	215A>G	–	Y/Y	–	Pol
	263A>G	–	Y/Y	rs2853515	Pol
	309.1insC	–	Y/Y	rs66492218	Pol/AD
	315.1insC	–	Y/Y	–	Pol
	318T>C	–	Y/Y	–	Pol
	326A>G	–	Y/Y	–	Pol
	489T>C	–	Y/Y	rs28625645	Pol
12S rRNA	750A>G	–	Y/Y	rs2853518	Pol
	1095T>C	–	Y/Y	rs267606618	Pol/SNHL
	1289G>A	–	Y/N	–	Pol
	1438A>G	–	Y/Y	rs2001030	Pol
16S rRNA	2706A>G	–	Y/Y	rs2854128	Pol
ND2	4769A>G	Syn(Met)	Y/Y	rs3021086	Pol
	5319A>G	Thr284Ala	Y/Y	rs28456039	Pol
CO1	6531C>T	Syn(Leu)	Y/Y	–	Pol
	7028C>T	Syn(Ala)	Y/Y	rs2015062	Pol
tRNA-Ser(UCN)	7471insC	–	Y/N	rs111033319	PEM/AMDF/motor neuron disease-like
CO2	7642G>A	Syn(Glu)	Y/Y	–	Pol
	8108A>G	Ile175Val	Y/Y	–	Pol/SNHL
ATP6	8701A>G	Thr59Ala	Y/Y	rs2000975	Pol
	8860A>G	Thr112Ala	Y/Y	rs2001031	Pol
CO3	9540T>C	Syn(Leu)	Y/Y	rs2248727	Pol
	9950T>C	Syn(Val)	Y/Y	rs3134801	Pol
ND3	10398A>G	Thr114Ala	Y/Y	rs2853826	Pol/PD/longevity
	10400C>T	Syn(Thr)	Y/Y	rs28358278	Pol
ND4	10873T>C	Syn(Pro)	Y/Y	rs2857284	Pol
	11719G>A	Syn(Gly)	Y/Y	rs2853495	Pol
	11969G>A	Ala404Thr	Y/Y	rs28359169	Pol
ND5	12705C>T	Syn(Ile)	Y/Y	rs2854122	Pol
	14053A>G	Thr573Ala	Y/Y	rs200134839	Pol
ND6	14502T>C	Ile58Val	Y/Y	rs201327354	Pol/LHON
CytB	14766C>T	Thr7Ile	Y/Y	rs3135031	Pol

Table 1. (*contd*)

Locus/gene	Mutation		Report		Polymorphism/phenotype[d]
	Nucleotide[a]	Amino acid[b]	Mitomap/mtDB[c]	dbSNP135	
	14783T>C	Syn(Leu)	Y/Y	rs28357680	Pol
	15043G>A	Syn(Gly)	Y/Y	rs28357684	Pol/MDD-associated
	15301G>A	Syn(Leu)	Y/Y	rs28573847	Pol
	15326A>G	Thr194Ala	Y/Y	rs2853508	Pol
D-loop	16169C>T	–	Y/Y	–	Pol
	16173C>T	–	Y/Y	–	Pol
	16192C>T	–	Y/Y	–	Pol
	16223C>T	–	Y/Y	rs2853513	Pol
	16243T>C	–	Y/Y	–	Pol
	16263T>C	–	Y/Y	–	Pol
	16292C>T	–	Y/Y	rs144417390	Pol
	16519T>C	–	Y/Y	rs3937033	Pol

The complete mtDNA sequence was registered in the GenBank database (KC709481). [a]Nt numbers are from the revised Cambridge reference sequence (NC_012920.1). [b]Syn, synonymous mutation; [c]Y, reported; N, unreported in the mtDB database or the MITOMAP database; [d]Pol, polymorphic; AD, Alzeimer's disease; SNHL, sensorineural hearing loss; PEM, progressive encephalopathy; PD, Parkinson's disease; LHON, Leber hereditary optic neuropathy; MDD, major depressive disorder.

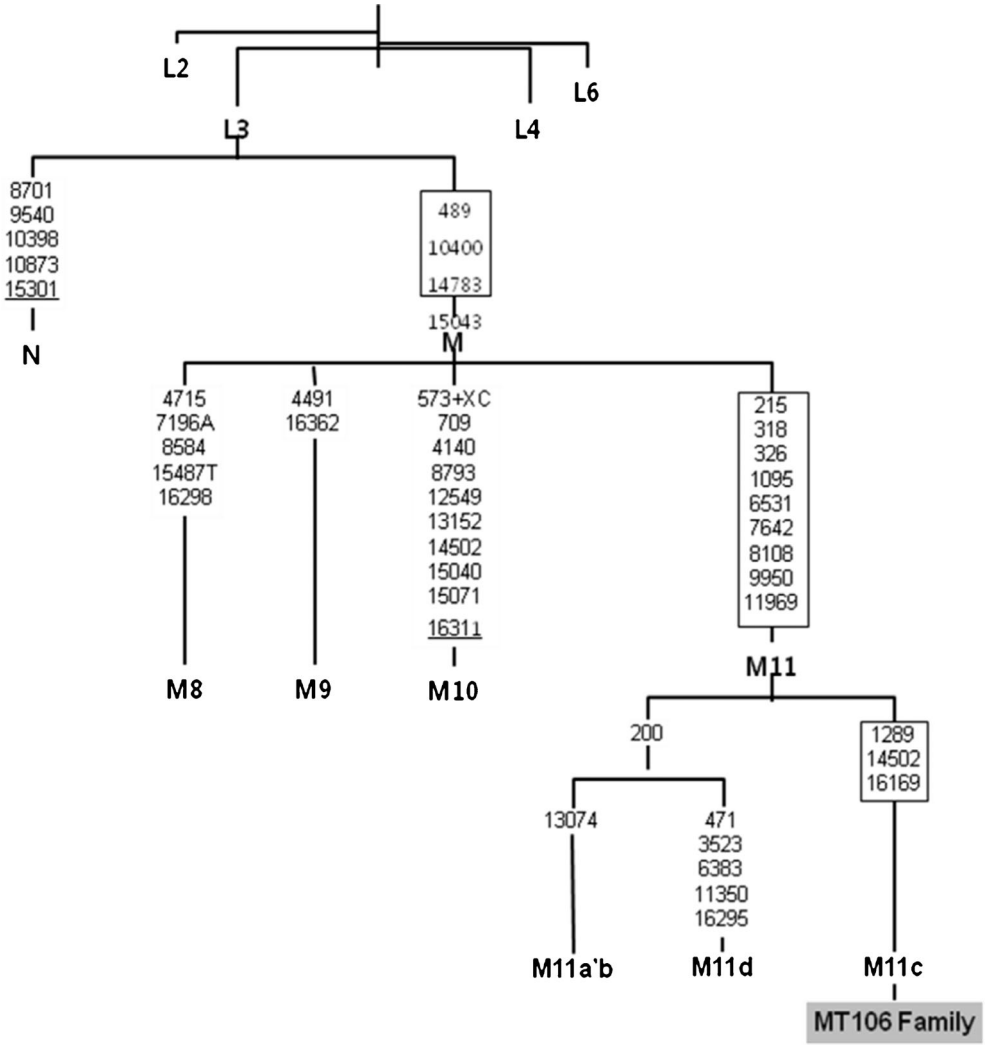

Figure 1. Determination of the mtDNA haplogroup. This haplogrouping was performed by the HaploGrep and the mtDNA manager programs. Recurrent mutations are underlined. They are transitions unless a base is explicitly indicated. Suffixes indicate transversions (to A, G, C, or T). The patient's mtDNA was determined to be the M11c group.