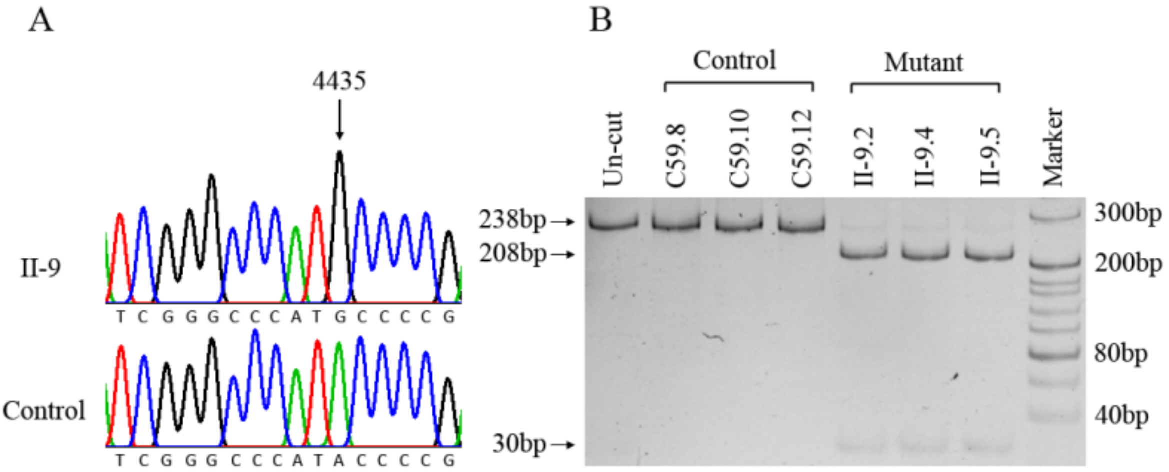
Supplemental Figure S1.



Supplemental Figure S1. Identification and quantification of m.4435A>G mutation in the tRNA$^{\mathrm{Met}}$ gene. (A) Partial sequence eletropherograms of the tRNA$^{\mathrm{Met}}$ gene from the an affected individual (II-9) and control subject (C59), respectively. (B) Quantification of the m.4435A>G mutation by PCR-RFLP. PCR products were digested with *N1aIII* and analyzed by electrophoresis in a 3% polyacrylamide gel stained with ethidium bromide. Patients and control cybrids are indicated.

Supplemental Table S1. Usage of methionine codons in mitochondrial proteins and the decrease level of mitochondrial proteins in the mutant cell lines related to the average value in the control cell lines

Proteins	Number of amino acids	Number of methionine codons	Density of methionine codons (%)	Decrease level of mitochondrial proteins (%)
ND3	115	8	7.0	58.5
ND4	459	27	5.9	8.3
ND5	604	26	4.3	2.5
COX2	225	10	4.4	22.9
CYTB	380	15	3.9	70.4
ATP6	227	12	5.3	49.4
ATP8	69	6	8.7	5.1
ND1	318	16	5.0	NA
ND2	347	25	7.2	NA
ND4L	99	10	10.1	NA
ND6	175	10	5.7	NA
COX1	514	32	6.2	NA
COX3	261	11	4.2	NA

NA, not applicable.

Supplemental Table S2. mtDNA variants in one Han Chinese hypertensive subject (II-9) and one control subject (C59)

Gene	Position	Replacement	Conservation (H/B/M/X)[a]	CRS[b]	II-9	C59	Previously reported[c]
D-loop	73	A to G		A	G	G	Yes
	152	T to C		T	G		Yes
	263	A to G		A	G	G	Yes
	310	T to CTC		T	CTC	CTC	Yes
	316	Ins C				ins C	N
	489	T to C		T	C	C	Yes
	16223	C to T		C	T	T	Yes
	16227	A to G		A	G		Yes
	16272	A to G		A	G		Yes
	16278	C to T		C	T		Yes
	16319	G to A		G	A	A	Yes
	16362	T to C		T	C	C	Yes
	16519	T to C		T	C		Yes
12S rRNA	709	G to A	G/A/A/-	G	A	A	Yes
	750	A to G	A/A/A/-	A	G	G	Yes
	1438	A to G	A/A/A/G	A	G	G	Yes
16S rRNA	2706	A to G	A/G/A/A	A	G	G	Yes
	3107	N to del	N/T/T/T	N	del N	del N	Yes
tRNA^{Met}	4435	A to G	A/A/A/A	A	G		Yes
ND2	4769			A	G	G	Yes
	4833	A to G(Thr to Ala)	T/I/I/L	A	G	G	Yes
	4973	T to C		T		C	Yes
	5108	T to C		T	C	C	Yes
CO1	7028	C to T		C	T	T	Yes
CO2	7600	G to A		G	A		Yes
	8161	C to T		C		T	Yes
	8200	T to C		T		C	Yes
ATP6	8701	A to G(Thr to Ala)	T/S/L/Q	A	G	G	Yes
	8860	A to G(Thr to Ala)	T/A/A/T	A	G	G	Yes
CO3	9377	A to G		A	G		Yes
	9540	T to C		T	C	C	Yes
	9575	G to A		G	A		Yes
	9966	G to A(Val to Ile)	V/V/V/V	G		A	Yes
ND3	10398	A to G(Thr to Ala)	T/T/T/A	A	G	G	Yes
	10400	C to T		C	T	T	Yes
ND4	10873	T to C		T	C	C	Yes
	11719	G to A		G	A	A	Yes
tRNA$^{Leu(CUN)}$	12280	A to G	A/G/A/A	A	G		Yes
ND5	12681	T to C		T		C	Yes

	12705	C to T		C	T	T	Yes
	13563	A to G		A	G		Yes
	14034	T to C		T	C	C	Yes
ND6	14569	G to A(Ser to Thr)	S/S/S/A	G	A	A	Yes
CYTB	14766	C to T(Thr to Ile)	T/S/T/S	C	T	T	Yes
	14783	T to C		T	C	C	Yes
	15043	G to A		G	A	A	Yes
	15301	G to A		G	A	A	Yes
	15323	G to A(Ala to Thr)	A/A/A/S	G		A	Yes
	15326	A to G(Thr to Ala)	T/M/I/I	A	G	G	Yes
	15497	G to A(Gly to Ser)	G/G/G/G	G		A	Yes

[a] Conservation of amino acid for polypeptides or nucleotide for RNAs in human (H), bovine (B), mouse (M) and *Xenopus laevis* (X).

[b] CRS: Cambridge reference sequence

[c] See the online mitochondrial genome database http://www.mi to map.org