

Table S1. mtDNA variants in three Han Chinese probands with hypertension and one control subject (C1)

Gene	Position	Replacement	Conservation		CRS	WHP20-II-13	WHP21-II-3	WHP22-II-5	C1	Previously reported ^b
			Index ^a	Frequency ^b						
			(17 species)							
D-loop	73	A to G			A	G	G	G	G	Yes
	150	C to T			C				T	Yes
	152	T to C			T		C			Yes
	195	T to C			T	G				Yes
	263	A to G			A	G	G	G	G	Yes
	298	C to T			C		T	T		Yes
	310	T to TC/TCC			T	TC	TCC	TCC	TC	Yes
	316	T to TC			T	TC	TC	TC		Yes
	489	T to C			T		C	C		Yes
	16093	T to C			T	C				Yes
	16136	T to C			T				C	Yes
	16140	T to C			T				C	Yes
	16150	C to T			C	T				Yes
	16169	C to T			C		T	T		Yes
	16182	A to C			A	C				Yes
	16183	A to C			A	C			C	Yes
	16189	T to C			T	C			C	Yes
	16193	C to CC			C				CC	Yes
	16217	T to C			T	C			C	Yes
	16223	C to T			C		T	T		Yes
	16240	A to G			A	G				Yes

	16249	T to C			T				C	Yes
	16261	C to T			C	T				Yes
	16274	G to A			G		A	A	A	Yes
	16280	A to G			A				G	Yes
	16291	C to T			C				T	Yes
	16311	T to C			T		C	C		Yes
	16362	T to C			T		C	C		Yes
	16407	C to T			C	C				Yes
	16519	T to C			T				C	Yes
12S									A	
rRNA	709	G to A	10/17	444/2704	G					Yes
	750	A to G	17/17	2682/2704	A	G	G	G	G	Yes
	1119	T to C	9/17	26/2704	T				C	Yes
	1438	A to G	16/17	2620/2704	A	G	G	G	G	Yes
	1556	C to T	4/17	0/2704	C	T				Yes
16S										
rRNA	1842	A to G	3/17	5/2704	A	G				Yes
	2483	T to C	6/17	7/2704	T				C	Yes
	2706	A to G	15/17	2178/2704	A	G			G	Yes
	3010	G to A	6/17	550/2704	G		A	A		Yes
<i>ND1</i>	3435	C to T			C				T	No
	3497	C to T (Ala64Val)	3/17	22/2704	C				T	Yes
	3571	C to T (Leu89Phe)	16/17	2/2704	C				T	No
	4140	C to T			C		T	T		No
tRNA ^{Gln}	4394	C to T	9/17	3/2704	C				T	Yes
<i>ND2</i>	4769	A to G			A		G	G		Yes

	4883	C to T			C		T	T	Yes
	5178	C to T (Leu237Met)	8/17	1/2704	C		A	A	Yes
tRNA^{Ala}	5655	T to C	10/17	23/2704	T	C	C	C	Yes
<i>COXI</i>	6286	T to C (Val128Ala)	17/17	1/2704	T	C			Yes
	6863	A to G			A	G			Yes
	7028	C to T			C	T	T	T	Yes
<i>NC7</i>	8281-8289	9bp del			C	9bp del		9 bp del	Yes
<i>ATP8</i>	8414	C to T (Leu17Phe)	10/17	264/2704	C		T	T	Yes
	8519	G to A (Glu52Lys)	12/17	1/2704	G		A	A	Yes
<i>ATP6</i>	8701	A to G (Thr59Ala)	9/17	933/2704	A		G	G	Yes
	8860	A to G (Thr112Ala)	12/17	2698/2704	A	G		G	Yes
<i>ATP6</i>	9128	T to C (Ile201Thr)		6/2704	T			C	Yes
<i>CO3</i>	9368	A to G			A		G	C	Yes
	9540	T to C			T		C	C	Yes
	9575	G to A			G			A	Yes
<i>ND3</i>	10398	A to G (Thr114Ala)	7/17	1242/2704	A	G	G	G	Yes
	10400	C to T (Thr114Ala)	7/17	724/2704	C		T	T	Yes
<i>ND4L</i>	10493	T to C			T			C	No
	10873	T to C			T		C	C	Yes
	11059	C to T			C		T	T	Yes
<i>ND4</i>	11440	G to A			G			A	Yes
	11719	G to A			G	A	A	A	Yes
<i>ND5</i>	12372	G to A			G		A	A	Yes
	12705	C to T			C		T	T	Yes
	13104	A to G			A		G	G	Yes
	13135	G to A (Ala267Ile)	4/17	18/2704	G	A			Yes

ND6	14410	G to A			G	A				Yes
	14668	C to T			C		T	T		Yes
Cytb	14766	C to T (Thr7Ile)	8/17	610/2704	C	T		T	T	Yes
	14783	T to C			T		C	C		Yes
	15043	G to A			G		A	A		Yes
	15085	A to G			A		G	G		Yes
	15301	G to A			G		A	A		Yes
tRNA ^{Thr}	15326	A to G (Thr194Ala)	9/17	2687/2704	A	G	G	G	G	Yes
	15924	T to C	14/17	106/2704	T			C		Yes

CRS, consensus Cambridge sequence (Andrews et al., 1999)

^a Conservation index (CI) was calculated by comparing the human mtDNA variants with other 16 vertebrates: *Bos Taurus*, *Cebus albifrons*, *Gorilla gorilla*, *Hylobates lar*, *Lemur catta*, *Macaca mulatta*, *Macaca sylvanus*, *Mus musculus*, *Nycticebus coucang*, *Pan paniscus*, *Pan troglodytes*, *Pongo pygmaeus*, *Pongo abelii*, *Papio hamadryas*, *Tarsius bancanus*, and *Xenopus laevis*.

^b See the online mitochondrial genome database <http://www.mitomap.org/MITOMAP> and <http://www.mtodb.igp.uu.se/>.

Table S2. Summary of Clinical Data for Some Members in three Chinese Pedigree

Subjects	Gender	Age of test (yrs)	Age of onset (yrs)	Diastolic pressure (mmHg)	Diastolic pressure (mmHg)
WHP20-I-2	F	94	80	180	90
WHP20-II-2	F	73	50	160	90
WHP20-II-3	M	69	64	170	80
WHP20-II-5	M	66	60	140	100
WHP20-II-6	F	62	48	150	110
WHP20-II-7	M	63	62	142	90
WHP20-II-9	M	60	55	160	110
WHP20-II-12	F	57	56	140	110
WHP20-II-13	M	55	50	210	130
WHP20-II-12	M	56	53	155	95
WHP20-III-1	M	55	40	180	110
WHP20-III-2	F	45	46	145	85
WHP20-III-3	M	52	52	140	90
WHP20-III-4	F	52	49	180	110
WHP20-III-6	F	48	42	170	110
WHP20-III-8	F	45	44	170	110
WHP20-III-20	M	40	-	110	70
WHP20-III-21	M	34	-	118	76
WHP20-IV-2	M	39	38	145	90

WHP20-IV-3	F	35	-	110	75
WHP20-IV-4	F	33	-	110	72
WHP20-IV-5	M	31	-	105	76
WHP20-IIV-6	F	28	-	114	70
WHP20-IIV-7	M	26	-	110	75
WHP21-I-2	F	75	-	135	78
WHP21-II-1	M	56	-	130	83
WHP21-II-3	F	55	55	145	98
WHP21-II-3	M	54	52	150	95
WHP21-II-8	F	48	-	125	79
WHP21-III-3	M	45	44	148	95
WHP21-III-4	M	44	-	120	75
WHP21-III-6	F	28	-	115	76
WHP21-III-7	F	26	-	112	70
WHP22-I-2	F	89	69	153	90
WHP22-II-1	F	69	64	165	110
WHP22-II-3	F	67	62	170	105
WHP22-II-5	F	65	60	158	101
WHP22-II-7	F	64	-	135	79
WHP22-II-9	M	63	59	156	115
WHP22-III-1	M	52	52	152	101
WHP22-III-2	M	51	-	135	79

WHP22-III-3	M	50	50	146	110
WHP22-III-4	F	49	-	138	84
WHP22-III-5	F	50	49	140	95
WHP22-III-6	F	48	-	138	82
WHP22-III-7	M	46	-	136	85
WHP22-III-8	F	48	-	130	79
WHP22-III-9	M	45	44	149	98
WHP22-III-10	M	44	-	130	76
WHP22-III-11	F	40	-	136	86

These patients had antihypertension treatment. This table shows pre-treatment blood pressures.