

Supplemental Table 1. mtDNA variants of the proband with maternally inherited HCM in this Han Chinese family

Gene	Position	Replacement	Conservation ^a (17 species)	Frequency ^b	CRS	Previously Reported ^b	Related Disease
D-loop	16223	C > T			C	Yes	
	16295	C > T			C	Yes	
	16519	T > C			T	Yes	
	73	A > G			A	Yes	
	146	T > C			T	Yes	
	199	T > C			T	Yes	
	489	T > C			T	Yes	
12S rRNA	750	A > G		2682/2704	A	Yes	
	1438	A > G		2620/2704	A	Yes	Diabetes ^c
16S rRNA	2336	T > C	16/17	0/2704	T	No	
	2706	A > G		2178/2704	A	Yes	
ND1	3882	G > A (syn)			G	Yes	
	4071	C > T (syn)			C	Yes	
ND2	4769	A > G (syn)			A	Yes	
	4850	C > T (syn)			C	Yes	
NC5(non-coding)	5442	T > C (Phe-Leu)	6/17	53/2704	T	Yes	
	5894	A > C	6/17	1/2704	A	Yes	
CO1	6455	C > T (syn)			C	Yes	
	7028	C > T (syn)			C	Yes	
ATP6	8701	A > G (Thr-Ala)	11/17	933/2704	A	Yes	lung cancer ^d
	8860	A > G (Thr-Ala)	10/17	2698/2704	A	Yes	
COIII	9266	G > A (syn)			G	Yes	
	9540	T > C (syn)			T	Yes	
	9824	T > C (syn)			T	Yes	

ND3	10398	A > G (Thr-Ala)	6/17	1242/2704	A	Yes	PD ^e
	10400	C > T (syn)			C	Yes	
ND4	10873	T > C (syn)			T	Yes	
	11665	C > T (syn)			C	Yes	
	11719	G > A (syn)			G	Yes	
	12091	T > C (syn)			T	Yes	
ND5	12372	G > A (syn)			G	Yes	
	12705	C > T (syn)			C	Yes	
	12810	A > G (syn)			A	Yes	
	13759	G > A (Ala-Thr)	1/17	39/2704	G	Yes	
Cytb	14766	C > T (Thr-Ile)	11/17	610/2704	C	Yes	
	14783	T > C (syn)			T	Yes	
	15043	G > A (syn)			G	Yes	
	15301	G > A (syn)			G	Yes	
	15326	A > G (Thr-Ala)	14/17	2687/2704	A	Yes	
	15364	C > T (syn)			C	No	

CRS, updated consensus Cambridge sequence (GenBank accession number: NC_012920)

^a Conservation of amino acid for polypeptides or nucleotide for RNAs in human and 16 organisms.

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

^c Yu Y et al. Mutations of mitochondrial 12S rRNA gene in type 2 diabetes. *Zhonghua Yi Xue Yi Chuan Xue Za Zhi*. 2001, 18:388-390.

^d Choi SJ et al. Mutational hotspots in the mitochondrial genome of lung cancer. *Biochem Biophys Res Commun*. 2011, 407:23-27.

^e Clark J et al. Association of PGC-1alpha polymorphisms with age of onset and risk of Parkinson's disease. *BMC Med Genet*. 2011, 12:69.