

**Supplemental Table S1. mtDNA variants in the proband (II-6) carrying the m.3635G>A mutation and two control subjects (C1,C2)**

Gene	Position	Replacement	Conservation Index (%) <sup>a</sup>	C1	C2	(II-6)	Previously reported <sup>b</sup>
D-loop	73	A-G		G	G	G	Yes
	143	G-A		A	A		Yes
	146	T-C				C	Yes
	150	C-T					Yes
	152	T-C		C		C	Yes
	185	G-A				A	Yes
	263	A-G		G	G	G	Yes
	309	C-CC					Yes
	310	T-TC/CTC		TC	TC	CTC	Yes
	489	T-C		C	C	C	Yes
	16188	C-T			T		Yes
	16189	T-C		C			Yes
	16223	C-T		T	T	T	Yes
	16227	A-G				G	Yes
	16262	C-T				T	Yes
	16274	G-A		A	A		Yes
	16278	C-T				T	Yes
	16294	C-T				T	Yes
	16319	G-A			A		Yes
	16362	T-C			C	C	C
12SrRNA	709	G-A	64.71	A	A	A	Yes
	750	A-G	100.00	G	G	G	Yes
	1438	A-G	94.12	G	G	G	Yes
16SrRNA	2706	A-G	100.00	G	G	G	Yes
<b>ND1</b>	<b>3635</b>	<b>G-A (Ser110Asn)</b>	<b>100.00</b>			<b>A</b>	<b>Yes</b>
	3882	G-A				A	Yes
ND2	4736	T-C		C			Yes
	4769	A-G		G	G	G	Yes
	4833	A-G (Thr122Ala)	47.06	G	G	G	Yes
	5093	T-C					Yes
	5108	T-C		C	C	C	Yes
	5237	G-A			G		Yes
	5471	G-A		A			Yes
Ala	5601	C-T	47.06			T	Yes
CO1	7028	C-T		T	T	T	Yes
CO2	7600	G-A			A	A	Yes
	7621	T-C		C	C		Yes
	7805	G-A (Val74Ile)	29.41			A	Yes
ATP6	8701	A-G (Thr59Ala)	52.94	G	G	G	Yes

	8860	A-G (Thr112Ala)	70.59	G	G	G	Yes
CO3	9377	A-G				G	Yes
	9540	T-C		C	C	C	Yes
	9575	G-A				A	Yes
ND3	10398	A-G (Thr114Ala)	41.18	G	G	G	Yes
	10400	C-T		T	T	T	Yes
ND4L	10609	T-C (Met47Thr)	52.94				Yes
ND4	10873	T-C		C	C	C	Yes
	11719	G-A		A	A	A	Yes
ND5	12705	C-T		T	T	T	Yes
	13563	A-G				G	Yes
ND6	14569	G-A		A	A	A	Yes
Cytb	14766	C-T (Thr7Ile)	47.06	T	T	T	Yes
	14783	T-C		C	C	C	Yes
	15043	G-A		A	A	A	Yes
	15301	G-A		A	A	A	Yes
	15326	A-G (Thr194Ala)	52.94	G	G	G	Yes
	15746	A-G (Ile334Val)		G	G		Yes

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<sup>a</sup> 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*.

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

**Supplemental Table S2. Summary of clinical and molecular data for members of one Chinese pedigree WZ513**

Subjects	Gender	Age of test (y)	Age of onset (y)	Vision acuity (Worst)		Level of vision Impairment	m.3635G>A mutation	YARS2 p.G191V mutation
				Right	Left			
II-1	M	62		0.8	0.7	Normal	+	+/+
II-2	F	58	17	0.1	0.1	Mild	-	+/-
II-3	M	55	16	0.04	0.02	Moderate	-	-/-
II-4	F	52		0.9	0.9	Normal	+	+/+
II-5	M	54		0.7	0.8	Normal	+	+/+
II-6	F	51	16	CF/20cm	CF/20cm	Profound	-	-/-
II-7	M	48		0.5	0.5	Normal	+	+/-
II-8	F	46		0.4	0.4	Normal	-	+/+
II-9	M	46	28	0.08	0.07	Severe	-	-/-
II-10	F	42		0.6	0.7	Normal	+	+/+
II-11	M	44		0.9	0.9	Normal	+	+/+
II-12	F	40		0.4	0.5	Normal	-	+/-
II-14	F	42	14	0.02	0.02	Severe	-	-/-
III-1	F	35		0.5	0.5	Normal	-	+/+
III-2	M	32		0.4	0.3	Normal	-	+/-
III-3	M	28		0.7	0.8	Normal	+	+/-
III-4	F	25		0.5	0.3	Normal	-	+/-
III-5	M	20		0.3	0.4	Normal	-	+/-
III-6	M	18		0.6	0.7	Normal	+	+/-
III-7	M	19		0.7	0.5	Normal	+	+/-
III-8	M	17		0.8	0.9	Normal	-	+/+

III-9	M	16		0.4	Normal	-	+/-	
III-10	F	17		0.7	0.8	Normal	-	+/+
III-11	F	18		0.6	0.8	Normal	-	+/+
III-12	M	16	16	0.01	0.02	Profound	-	-/+

The degree of visual impairment was defined according to the visual acuity as follows: normal >0.3, mild =0.3-0.1; moderate<0.1-0.05; severe<0.05-0.02; and profound <0.02.

M, Male; F, Female; CF, counting fingers;