

Table 3S. Rare mtDNA mutations associated with diseases and their distribution in the 90+ and controls of the present study.

Position	Mutation type	Controls		90+		Region	Disease
		N.	%	N.	%		
827	A/G	1	0,15	0	0,00	12S	Deafness
961	T/C	2	0,31	5	0,77	12S	Deafness
1005	T/C	1	0,15	0	0,00	12S	Deafness
1438	A/G	25	3,87	39	6,04	12S	Diabetes Mellitus
1555	A/G	0	0,00	3	0,46	12S	Deafness
3196	G/A	1	0,15	0	0,00	16S	Alzheimer & Parkinson Disease
3308	T/C	1	0,15	0	0,00	ND1	Encephalomyopathy, MELAS
3460	G/T	0	0,00	0	0,00	ND1	LHON
3796	A/G	3	0,46	5	0,77	ND1	Dystonia
4295	A/G	8	1,24	4	0,62	TI	Maternally Inherited Hypertrophic Cardiomyopathy
4336	T/C	5	0,77	12	1,86	TQ	Alzheimer & Parkinson Disease Deafness & Migraine
5460	G/A	36	5,57	31	4,80	ND2	Alzheimer & Parkinson Disease
5628	T/C	1	0,15	0	0,00	tRNA A	Mitochondrial Myopathy, CPEO MTTA CPEO
5843	A/G	1	0,15	0	0,00	TY	Mitochondrial Myopathy Cytopathy
6489	C/A	5	0,77	2	0,31	CO1	Epilepsy
7444	G/A	4	0,62	6	0,93	CO1	Deafness-Sensory Neural Hearing Loss
8108	A/G	0	0,00	1	0,15	CO1	Deafness-Sensory Neural Hearing Loss

8302	A/G	0	0,00	1	0,15	tRNA k	MTTK Encephalopathy
8993	T/C	0	0,00	0	0,00	ATP6	Leigh Syndrome
10044	A/G	8	1,24	3	0,46	TG	Encephalomyopathy Gastrointestinal Reflux and Sudden Infant Death Syndrome
11084	A/G	1	0,15	0	0,00	ND4	Encephalomyopathy, MELAS
12026	A/G	0	0,00	2	0,31	ND4	Diabetes Mellitus
12192	G/A	0	0,00	3	0,46	TH	Maternally Inherited Cardiomyopathy
12308	A/G	149	23,07	116	17,96	TL2	Mitochondrial Myopathy, CPEO, Stroke
15497	G/A	3	0,46	1	0,15	CYTB	MM, Exercise Intolerance
15498	G/A	0	0,00	1	0,15	CYTB	Hypertrophic Cardiomyopathy
15924	A/G	17	2,63	27	4,18	TT	Mitochondrial Myopathy Lethal Infantile Mitochondrial Myopathy

Table 4S: Distribution of 90+ and controls from different centers participating to GEHA project.

Recruitment center	Country	No. 90+	No. younger controls
Belfast	Northern Ireland	64	64
Louvain	Belgium	80	88
Bologna	Italy	213	214
Kiel	Germany	94	96
Tampere	Finland	153	145
Montpellier	France	274	275
Newcastle	UK	99	100
Odense	Denmark	428	441
Leiden	Netherland	162	167
Warsaw	Poland	129	132
Rome	Italy	75	75
Sassari	Italy	47	52
Kiev	Ukraina	49	49
Calabria	Italy	125	152
Athens	Greece	94	103
Total		2,086	2,153

Table 5S: Sample distribution and general characteristics of the study participants.

	Males (N = 1,327)		Females (N = 2,912)	
	90+ (%)	Controls (%)	90+ (%)	Controls (%)
N	628 (47)	699 (53)	1,458 (50)	1,454 (50)
Mean Age	94,1 ± 2,5	61,7 ± 6,2	94,5 ± 2,6	61,7 ± 6,2
Age range	89-103	43-79	88-107	49-83

Table 6S: Modified primer pairs for the complete resequencing (M13 tag is not reported).

	Forward	Reverse
Primer 41b	5'- CTG CTT CTT CTC ACA TGA-3'	5'- TAG GTA GGA GTA GCG TG-3'
Primer 43b	5'- ATC AAT TGG CTT CCT AGG-3'	5'- GGC ATC CAT ATA GTC ACT-3'
Primer 44b	5'- CCT TAT CTG CTT CCT AGT-3'	5'-GGG TGC TAT AGG GTA AAT -3'
Primer 45b	5'- ATT TTG ACT ACC ACA ACT CA-3'	5'-ATA TGT GTT GGA GAT TGA GA -3'