

Appendix 3. List of complete mtDNA variants observed in Family B.

Nucleotide Position	rCRS	Base change	Location	Codon	Position	Amino acid Change	Synonymous	Significance
73	A	G	D-loop					
152	T	C	D-loop					
217	T	C	D-loop					
263	A	C	D-loop					
310		CC INS	D-loop					
316		C INS	D-loop					
340	C	T						
508	A	G	D-loop					
750	A	G	12S rRNA					
988	G	A	12S rRNA					
1438	A	G	12S rRNA					
1555	A	G	12S rRNA					DEAF
1811	A	G	16S rRNA					
2706	A	G	16S rRNA					
3720	A	G	ND1	138	3	Gln -> Gln	Yes	
4769	A	G	ND2	100	3	Met -> Met	Yes	
5390	A	G	ND2	307	3	Met -> Met	Yes	
5426	T	C	ND2	319	3	His -> His	Yes	
5899		CC INS						
6045	C	T	COI	48	1	Leu -> Leu	Yes	
6152	T	C	COI	83	3	Val -> Val	Yes	
7028	C	T	COI	375	3	Ala -> Ala	Yes	
8860	A	G	ATPase6	112	1	Thr -> Ala	No	
10876	A	G	ND4	39	3	Leu -> Leu	Yes	
11467	A	G	ND4	236	3	Leu -> Leu	Yes	
11719	G	A	ND4	320	3	Gly -> Gly	Yes	
11778	G	A	ND4	340	2	Arg -> His	No	LHON
12308	A	G	tRNA Leu					
12372	G	A	ND5	12	3	Leu -> Leu	Yes	
13020	T	C	ND5	228	3	Gly -> Gly	Yes	
13734	T	C	ND5	466	3	Phe -> Phe	Yes	
14766	T	C	Cytb	7	2	Ile -> Thr	No	
15326	A	G	Cytb	194	1	Thr -> Ala	No	
15907	A	G	tRNA Thr					
16051	A	G	D-loop					
16092	T	C	D-loop					
16129	G	C	D-loop					
16189	T	C	D-loop					
16362	T	C	D-loop					