

Appendix 2. List of complete mtDNA variants observed in Family A.

Nucleotide Position	rCRS	Base Change	Location	Codon	Position	Amino acid Change	Synonymous	Significance
73	A	G	D-loop					
152	T	C	D-loop					
263	A	C	D-loop					
316		C INS	D-loop					
489	T	C	D-loop					
494	C	T	D-loop					
709	G	A	D-loop					
750	A	G	12S rRNA					
1438	A	G	12S rRNA					
1555	A	G	12S rRNA					DEAF
1888	G	A	16S rRNA					
2706	A	G	16S rRNA					
3921	C	T	ND1	205	3	Ser -> Ser	Yes	
4769	A	G	ND2	100	3	Met -> Met	Yes	
6047	A	G	COI	48	3	Leu -> Leu	Yes	
7028	C	T	COI	375	3	Ala -> Ala	Yes	
8020	G	A	COII	145	3	Pro -> Pro	Yes	
8701	A	G	ATPase6	59	1	Thr -> Ala	No	
8860	A	G	ATPase6	112	1	Thr -> Ala	No	
9540	T	C	COIII	112	1	Leu -> Leu	Yes	
10398	A	G	ND3	114	1	Thr -> Ala	No	
10400	C	T	ND3	114	3	Thr -> Ala	No	
10873	T	C	ND4	38	3	Pro -> Pro	Yes	
11719	G	A	ND4	320	3	Gly -> Gly	Yes	
11778	G	A	ND4	340	2	Arg -> His	No	LHON
12477	T	C	ND5	47	3	Ser -> Ser	Yes	
12705	C	T	ND5	123	3	Ile -> Ile	Yes	
14323	G	A	ND6	117	3	Asn -> Asn	Yes	
14766	T	C	Cytb	7	2	Ile -> Thr	No	
14783	T	C	Cytb	13	1	Leu -> Leu	Yes	
15043	G	A	Cytb	99	3	Gly -> Gly	Yes	
15301	G	A	Cytb	185	3	Leu -> Leu	Yes	
15326	A	G	Cytb	194	1	Thr -> Ala	No	
16093	T	C	D-loop					
16129	G	C	D-loop					
16223	C	T	D-loop					