

Patient ID		rCRS	P1	P2	P3	P4	P5	P6	P7	P8	P9	P10	P11	P12	P13	P14	P15	P16	P17	P18	P19	P20	Location	Amino acid	Significance	Frequency
Nucleotide position	150	C													T			T					D-Loop	NA	Longevity/Cervical carcinoma/HPV infection risk	2/20
	195	T					C																D-Loop	NA	BD associated	1/20
	309	C	insC	insCC						insC	insCC		insC				insC	insC			insC	insC	D-Loop	NA	AD weakly associated	9/20
	5460	G																				A	ND2	H319H	AD/PD	1/20
	7637	G					A																CO2	E18K	PD risk factor	1/20
	10398	A		G	G		G	G			G	G		G				G					ND3	T114A	PD protective factor/longevity/altered cell pH/metabolic syndrome/breast cancer risk/IJK haplogroup	8/20
	11026	A																G					ND4	P89P	Novel	1/20
	11467	A	G						G	G					G								ND4	L236L	Altered brain pH	4/20
	12330	A									G												tRNA Leu	NA	Novel	1/20
	12372	G	A						A	A					A								ND5	L12L	Altered brain pH	4/20
	13135	G						A				A											ND5	A267T	Possible HCM susceptibility	2/20
	13392	T										C											ND5	L286L	Novel	1/20
	13708	G										A						A					ND5	A458T	LHON/increased MH risk/high frequency in PDADS/ haplogroup J marker	2/20
	16519	T	C	C	C		C	C			C			C			C	C	C	C	C	C	D-Loop	NA	Cyclic Vomiting syndrome with migraine	13/20
Haplogroup			<i>H1a3</i>	<i>M3c1a</i>	<i>M3a1</i>	<i>H2a2a</i>	<i>H2a2a</i>	<i>L3*</i>	<i>U2b</i>	<i>U2</i>	<i>H2a2a</i>	<i>H2a2a</i>	<i>H2a2a</i>	<i>M49c</i>	<i>U5b2a1a</i>	<i>H2a2a</i>	<i>H2a2a</i>	<i>M5c1</i>	<i>R31a1</i>	<i>H2a2a</i>	<i>H2a2a</i>	<i>H2a2a</i>				

Appendix 2: Disease associated mtDNA nucleotide changes observed in the PCG patients, their significance and haplogroups.
(rCRS – revised Cambridge Reference Sequence)