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Supplementary Information for

Biparental Inheritance of Mitochondrial DNA in Humans

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Tables S1-S3

Other supplementary materials for this manuscript include the following:

Datasets S1 to S3

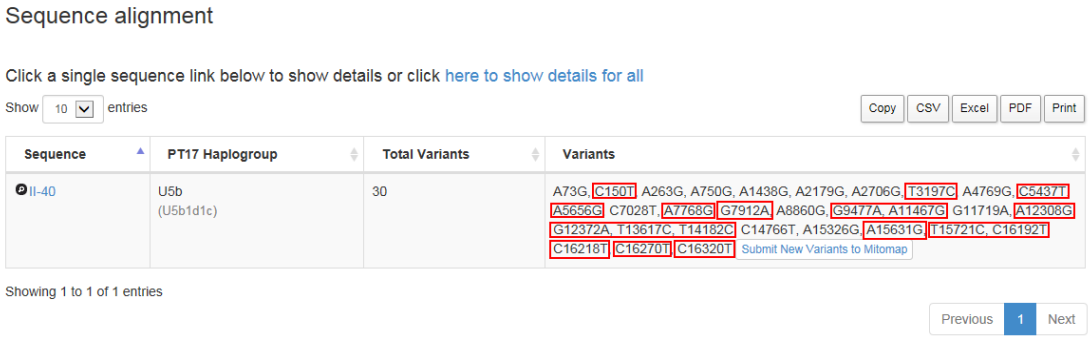

Fig. S1. The 30 SNPs in II-40's mtDNA (Family A) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup U5b (U5b1d1c)-related mtDNA variants are labeled in red rectangles.

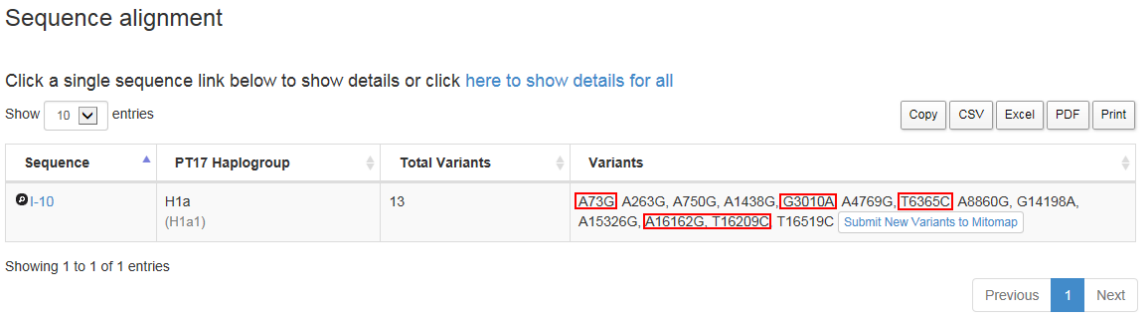

Fig. S2. The 13 SNPs in I-10's mtDNA (Family A) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup H1a (H1a1)-related mtDNA variants are labeled in red rectangles in addition to other defining SNPs (2706A, 7028C).

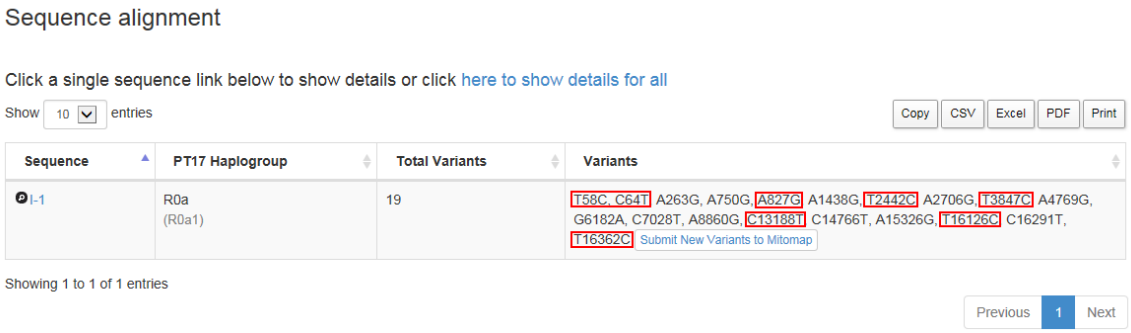

Fig. S3. The 19 SNPs in I-1's mtDNA (Family A) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup R0a (R0a1)-related mtDNA variants are labeled in red rectangles.

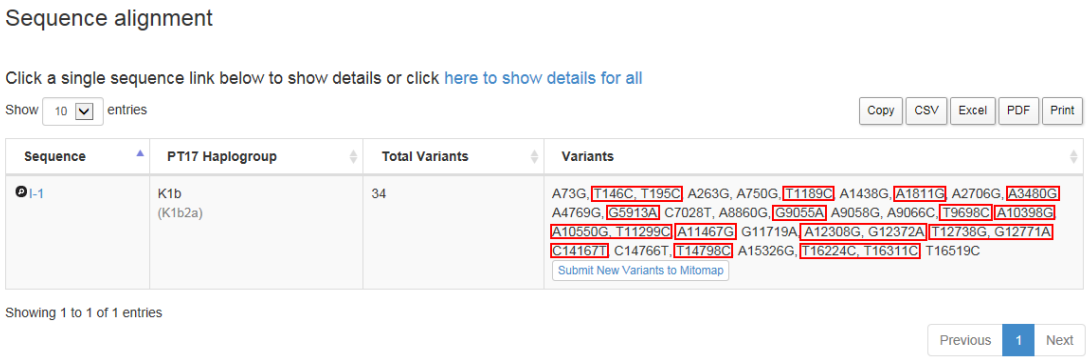

Fig. S4. The 34 SNPs in I-1's mtDNA (Family B) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup K1b (K1b2a)-related mtDNA variants are labeled in red rectangles.

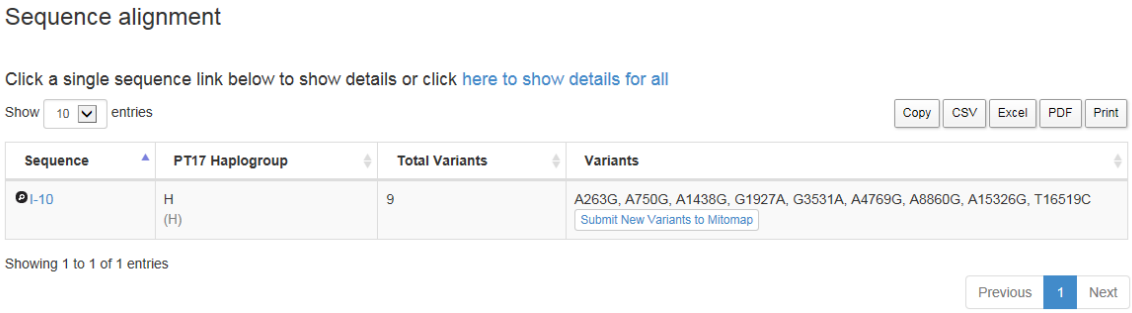

Fig. S5. The nine SNPs in I-10's mtDNA (Family B) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup H is defined by mtDNA genotype of 73A, 2706A, 7028C, 11719G and 14766C.

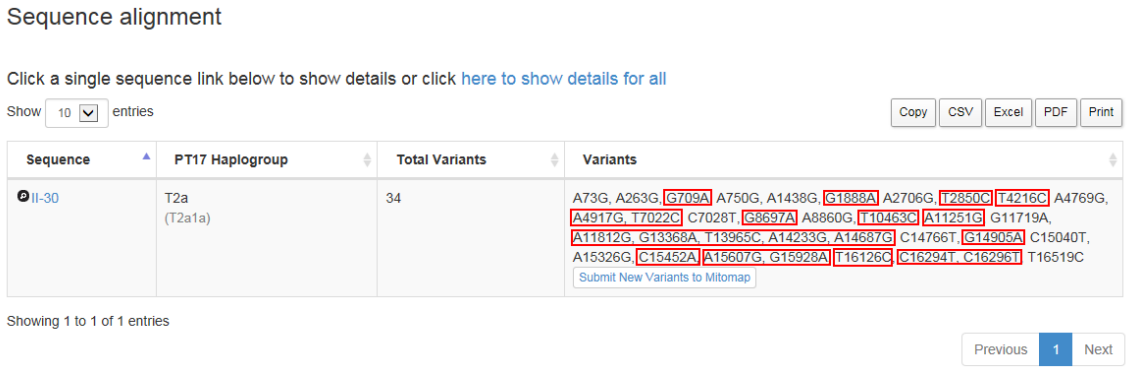

Fig. S6. The 34 SNPs in II-30's mtDNA (Family B) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup T2a (T2a1a)-related mtDNA variants are labeled in red rectangle.

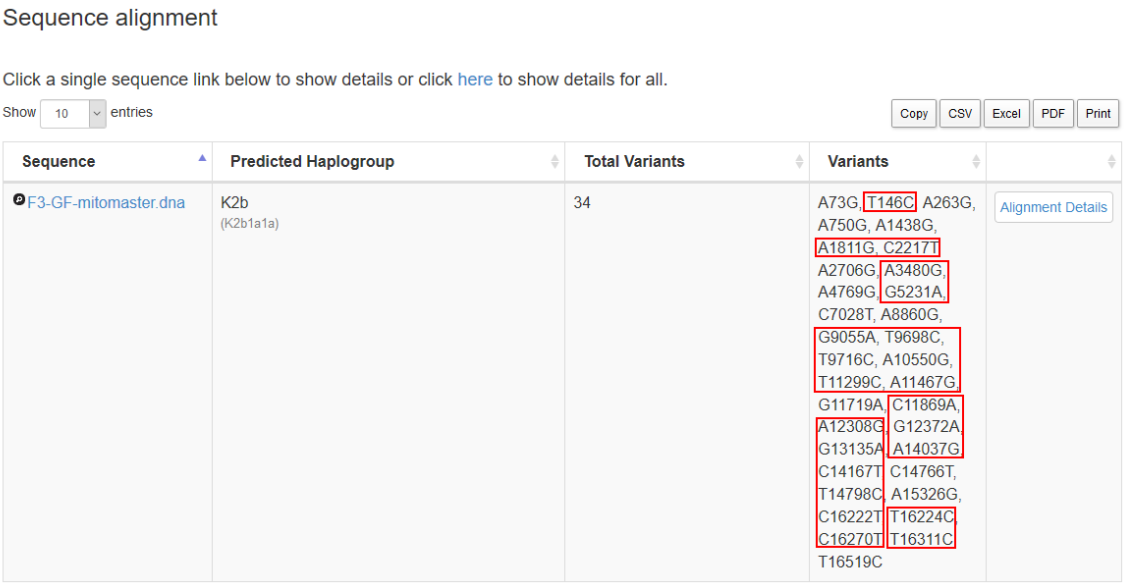

Fig. S7. The 34 SNPs in I-1's mtDNA (Family C) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup K2b (K2b1a1a)-related mtDNA variants are labeled in red rectangle.

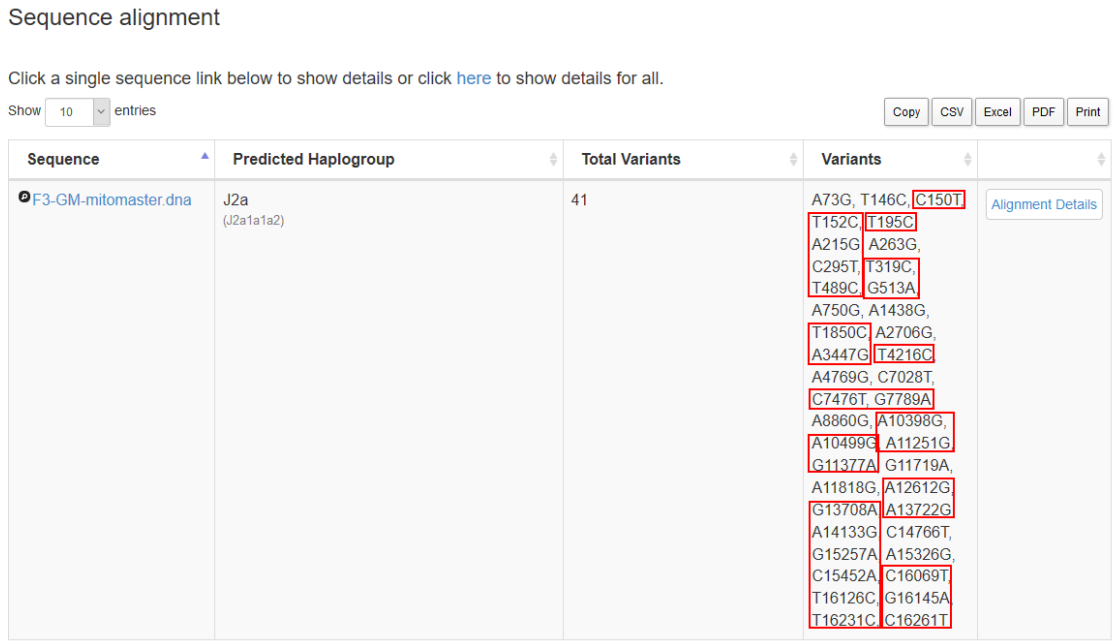

Fig. S8. The 41 SNPs in I-10's mtDNA (Family C) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup J2a (J2a1a1a2)-related mtDNA variants are labeled in red rectangle.

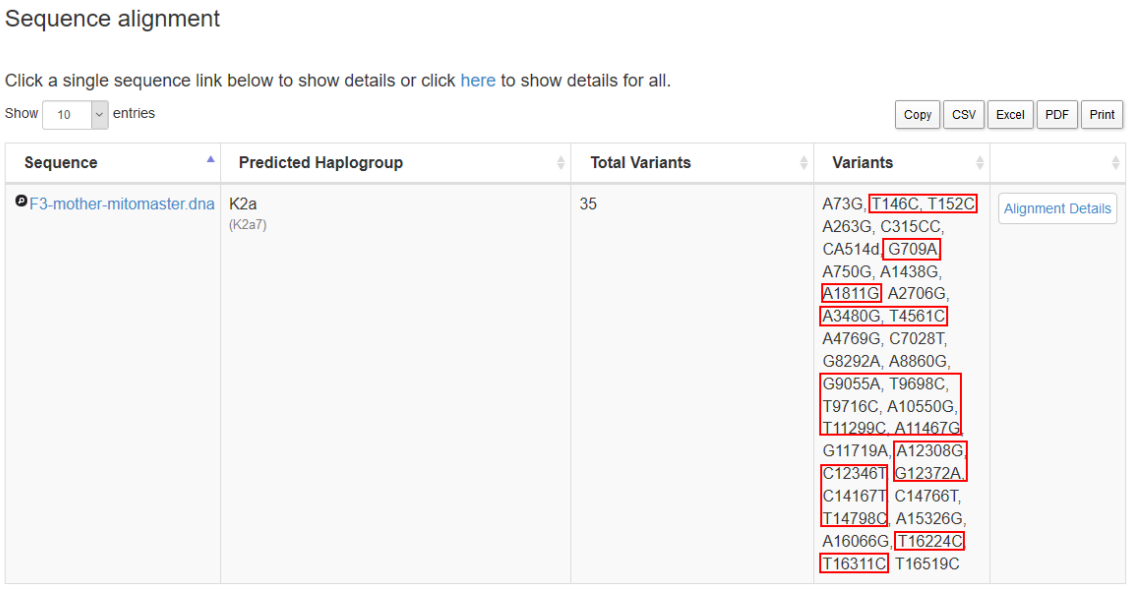

Fig. S9. The 35 SNPs in II-30's mtDNA (Family C) differing from the reference mtDNA haplogroup were predicted in Mitomaster. Haplogroup K2a (K2a7)-related mtDNA variants are labeled in red rectangle.

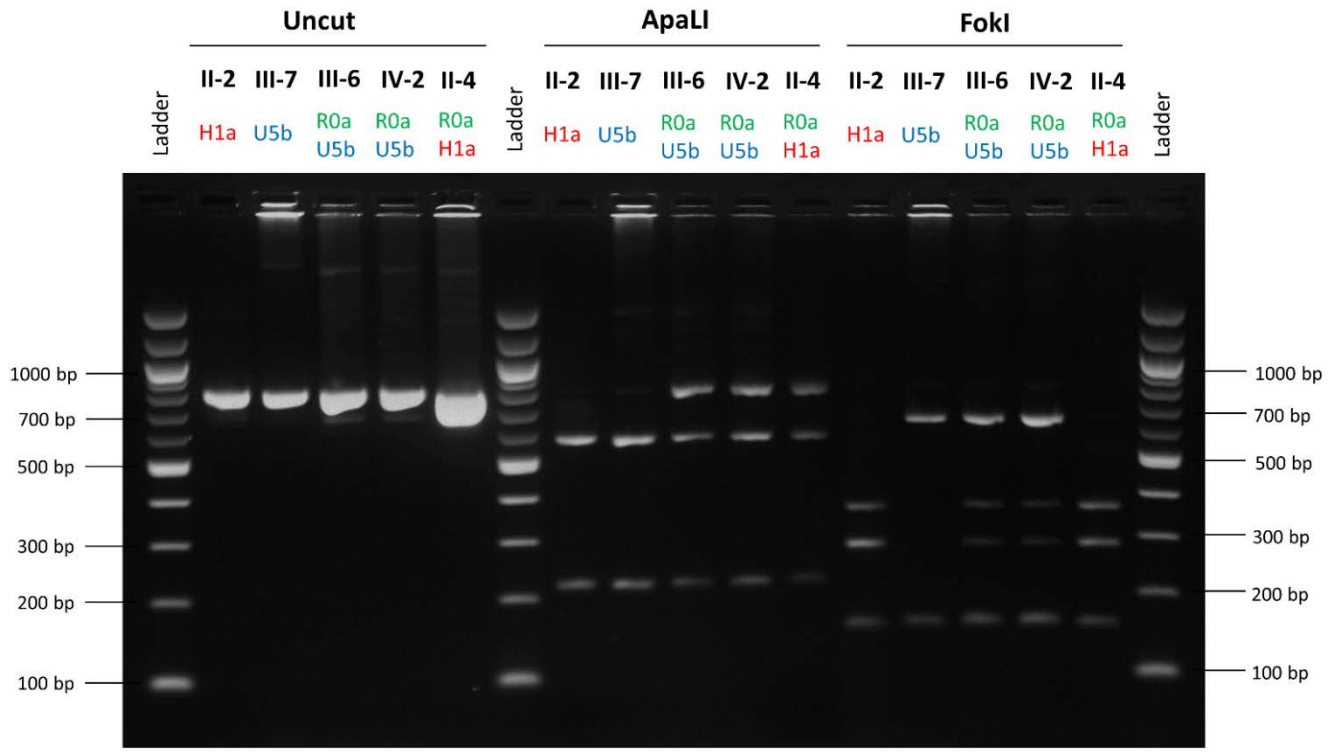

Fig. S10. RFLP analysis of selected members of Family A confirms the existence of the haplogroups detected by NGS. The haplogroups present in each individual are as indicated ("R0a" = R0a1, "H1a" = H1a1, and "U5b" = U5b1d1c). RFLP was performed on a 789 bp

PCR product covering positions 16426 to 626 in the human mitochondrial reference genome, which includes the hypervariable region HV2. The expected digest pattern for ApaLI is 789 bp for R0a1 and 572 bp and 217 bp for H1a1 and U5b1d1c. For FokI, the expected digest pattern is 632 bp and 157 bp for U5b1d1c and 355 bp, 277 bp, and 157 bp for R0a1 and H1a1.

Dataset S1. Comparison of mtDNA sequences of 11 family members in Family A. The mtDNA sequences were aligned with reference sequence- the human mitochondrial sequence reference NC_012920.1. SNPs were identified and submitted to MitoMaster for variant calls. All de novo SNPs were labeled with gray color and not included for analysis of mtDNA inheritance pattern in this family.

Dataset S2. Comparison of mtDNA sequences of five family members in Family B. The mtDNA sequences were aligned with reference sequence- the human mitochondrial sequence reference NC_012920.1. SNPs were identified and submitted to MitoMaster for variant calls.

Dataset S3. Comparison of mtDNA sequences of five family members in Family C. The mtDNA sequences were aligned with reference sequence- the human mitochondrial sequence reference NC_012920.1. SNPs were identified and submitted to MitoMaster for variant calls.

Table S1. Alignment of a total of 46 differential mtDNA variants in family A

rCRS Position	Reference	Variant	I-1 (D)	I-10 (D)/II-2/III-3	II-1/II-3/II-4	II-40/III-7	III-6/IV-1/IV-2/IV-3
58	T	C	C	T	C/T	T	C/T
64	C	T	T	C	T/C	C	T/C
73	A	G	A	G	A/G	G	A/G
150	C	T	C	C	C	T	C/T
263	A	G	G	G	G	G	G
750	A	G	G	G	G	G	G
827	A	G	G	A	G/A	A	G/A
1438	A	G	G	G	G	G	G
2179	A	G	A	A	A	G	A/G
2442	T	C	C	T	C/T	T	C/T
2706	A	G	G	A	G/A	G	G
3010	G	A	G	A	G/A	G	G
3197	T	C	T	T	T	C	T/C
3847	T	C	C	T	C/T	T	C/T
4769	A	G	G	G	G	G	G
5437	C	T	C	C	C	T	C/T
5656	A	G	A	A	A	G	A/G
6182	G	A	A	G	A/G	G	A/G
6365	T	C	T	C	T/C	T	T
7028	C	T	T	C	T/C	T	T
7768	A	G	A	A	A	G	A/G
7912	G	A	G	G	G	A	G/A
8860	A	G	G	G	G	G	G
9477	G	A	G	G	G	A	G/A
11467	A	G	A	A	A	G	A/G
11719	G	A	G	G	G	A	G/A
12308	A	G	A	A	A	G	A/G
12372	G	A	G	G	G	A	G/A
13188	C	T	T	C	T/C	C	T/C
13617	T	C	T	T	T	C	T/C
14182	T	C	T	T	T	C	T/C
14198	G	A	G	A	G/A	G	G
14766	C	T	T	C	T/C	T	T
15326	A	G	G	G	G	G	G
15631	A	G	A	A	A	G	A/G
15721	T	C	T	T	T	C	T/C
16126	T	C	C	T	C/T	T	C/T
16162	A	G	A	G	A/G	A	A
16192	C	T	C	C	C	T	C/T
16209	T	C	T	C	T/C	T	T
16218	C	T	C	C	C	T	C/T
16270	C	T	C	C	C	T	C/T
16291	C	T	T	C	T/C	C	T/C
16320	C	T	C	C	C	T	C/T
16362	T	C	C	T	C/T	T	C/T
16519	T	C	T	C	T/C	T	T
Heteroplasmy %			100	100	40/60	100	40/60
Haplogroup			R0a1	H1a1	R0a1/H1a1	U5b1d1c	R0a1/U5b1d1c

Entries labeled (D) represent deduced mtDNA genotypes. Haplogroup related nucleotides have been underlined in the table.

Table S2. Alignment of a total of 58 differential mtDNA variants in family B

rCRS Position	Reference	Variant	I-1 (D)	I-10 (D)	II-3	II-30/III-1/III-5	III-2
73	A	G	G	A	G/A	G	G
146	T	C	C	T	C/T	T	C/T
195	T	C	C	T	C/T	T	C/T
263	A	G	G	G	G	G	G
709	G	A	G	G	G	A	G/A
750	A	G	G	G	G	G	G
1189	T	C	C	T	C/T	T	C/T
1438	A	G	G	G	G	G	G
1811	A	G	G	A	G/A	A	G/A
1888	G	A	G	G	G	A	G/A
1927	G	A	G	A	G/A	G	G
2706	A	G	G	A	G/A	G	G
2850	T	C	T	T	T	C	T/C
3480	A	G	G	A	G/A	A	G/A
3531	G	A	G	A	G/A	G	G
4216	T	C	T	T	T	C	T/C
4769	A	G	G	G	G	G	G
4917	A	G	A	A	A	G	A/G
5913	G	A	A	G	A/G	G	A/G
7022	T	C	T	T	T	C	T/C
7028	C	T	T	C	T/C	T	T
8697	G	A	G	G	G	A	G/A
8860	A	G	G	G	G	G	G
9055	G	A	A	G	A/G	G	A/G
9058	A	G	G	A	G/A	A	G/A
9066	A	C	C	A	C/A	A	C/A
9698	T	C	C	T	C/T	T	C/T
10398	A	G	G	A	G/A	A	G/A
10463	T	C	T	T	T	C	T/C
10550	A	G	G	A	G/A	A	G/A
11251	A	G	A	A	A	G	A/G
11299	T	C	C	T	C/T	T	C/T
11467	A	G	G	A	G/A	A	G/A
11719	G	A	A	G	A/G	A	A
11812	A	G	A	A	A	G	A/G
12308	A	G	G	A	G/A	A	G/A
12372	G	A	A	G	A/G	G	A/G
12738	T	G	G	T	G/T	T	G/T
12771	G	A	A	G	A/G	G	A/G
13368	G	A	G	G	G	A	G/A
13965	T	C	T	T	T	C	T/C
14167	C	T	T	C	T/C	C	T/C
14233	A	G	A	A	A	G	A/G
14687	A	G	A	A	A	G	A/G
14766	C	T	T	C	T/C	T	T
14798	T	C	C	T	C/T	T	C/T
14905	G	A	G	G	G	A	G/A
15040	C	T	C	C	C	T	C/T
15326	A	G	G	G	G	G	G
15452	C	A	C	C	C	A	C/A
15607	A	G	A	A	A	G	A/G
15928	G	A	G	G	G	A	G/A
16126	T	C	T	T	T	C	T/C
16224	T	C	C	T	C/T	T	C/T
16294	C	T	C	C	C	T	C/T
16296	C	T	C	C	C	T	C/T
16311	T	C	C	T	C/T	T	C/T
16519	T	C	C	C	C	C	C
Heteroplasmy %			100	100	28/72	100	51/49
Haplogroup			K1b2a	H	K1b2a/H	T2a1a	K1b2a/T2a1a

Entries labeled (D) represent deduced mtDNA genotypes. Haplogroup related nucleotides have been underlined in the table.

Table S3. Alignment of a total of 68 differential mtDNA variants in family C

rCRS Position	Reference	Variant	I-1 (D)	I-10 (D)	II-3	II-30	III-6/III-7/IV-1	rCRS Position	Reference	Variant	I-1 (D)	I-10 (D)	II-3	II-30	III-6/III-7/IV-1
73	A	G	G	G	G	G	G	10550	A	G	G	A	G/A	G	G
146	T	C	C	C	C	C	C	11251	A	G	A	G	A/G	A	A
150	C	T	C	T	C/T	C	C	11299	T	C	C	T	C/T	C	C
152	T	C	T	C	T/C	C	T/C	11377	G	A	G	A	G/A	G	G
195	T	C	T	C	T/C	T	T	11467	A	G	G	A	G/A	G	G
215	A	G	A	G	A/G	A	A	11719	G	A	A	A	A	A	A
263	A	G	G	G	G	G	G	11818	A	G	A	G	A/G	A	A
295	C	T	C	T	C/T	C	C	11869	C	A	A	C	A/C	C	A/C
319	T	C	T	C	T/C	T	T	12308	A	G	G	A	G/A	G	G
489	T	C	T	C	T/C	T	T	12346	C	T	C	C	C	T	C/T
513	G	A	G	A	G/A	G	G	12372	G	A	A	G	A/G	A	A
709	G	A	G	G	G	A	G/A	12612	A	G	A	G	A/G	A	A
750	A	G	G	G	G	G	G	13135	G	A	A	G	A/G	G	A/G
1438	A	G	G	G	G	G	G	13708	G	A	G	A	G/A	G	G
1811	A	G	G	A	G/A	G	G	13722	A	G	A	G	A/G	A	A
1850	T	C	T	C	T/C	T	T	14037	A	G	G	A	G/A	A	G/A
2217	C	T	T	C	T/C	C	T/C	14133	A	G	A	G	A/G	A	A
2706	A	G	G	G	G	G	G	14167	C	T	T	C	T/C	T	T
3447	A	G	A	G	A/G	A	A	14766	C	T	T	T	T	T	T
3480	A	G	G	A	G/A	G	G	14798	T	C	C	T	C/T	C	C
4216	T	C	T	C	T/C	T	T	15257	G	A	G	A	G/A	G	G
4561	T	C	T	T	T	C	T/C	15326	A	G	G	G	G	G	G
4769	A	G	G	G	G	G	G	15452	C	A	C	A	C/A	C	C
5231	G	A	A	G	A/G	G	A/G	16066	A	G	A	A	A	G	A/G
7028	C	T	T	T	T	T	T	16069	C	T	C	T	C/T	C	C
7476	C	T	C	T	C/T	C	C	16126	T	C	T	C	T/C	T	T
7789	G	A	G	A	G/A	G	G	16145	G	A	G	A	G/A	G	G
8292	G	A	G	G	G	A	G/A	16222	C	T	T	C	T/C	C	T/C
8860	A	G	G	G	G	G	G	16224	T	C	C	T	C/T	C	C
9055	G	A	A	G	A/G	A	A	16231	T	C	T	C	T/C	T	T
9698	T	C	C	T	C/T	C	C	16261	C	T	C	T	C/T	C	C
9716	T	C	C	T	C/T	C	C	16270	C	T	T	C	T/C	C	T/C
10398	A	G	A	G	A/G	A	A	16311	T	C	C	T	C/T	C	C
10499	A	G	A	G	A/G	A	A	16519	T	C	C	T	C/T	C	C
Heteroplasmy %			100	100	70/30	100	76/24	Heteroplasmy %			100	100	70/30	100	76/24
Haplogroup			K2b1a1a	J2a1a1a2	K2b1a1a/J2a1a1a2	K2a7	K2b1a1a/K2a7	Haplogroup			K2b1a1a	J2a1a1a2	K2b1a1a/J2a1a1a2	K2a7	K2b1a1a/K2a7

Entries labeled (D) represent deduced mtDNA genotypes. Haplogroup related nucleotides have been underlined in the table.