Table S1 Correlation of the number of nonaggregate haplotypes by nucleotide substitutions per 100 single cells with age

| Samples | r | $r^2$ | *P*-value |
|---|---|---|---|
| All 51 samples from 49 subjects | 0.44 | 0.19 | 0.001 |
| All samples excluding the three cord blood samples and one adolescent sample | 0.41 | 0.17 | 0.004 |
| All samples (controlling for family)[1] | 0.40 | — | 0.004 |
| All samples excluding the three cord blood samples and one adolescent sample (controlling for family)[1] | 0.40 | — | 0.007 |
| Family A only | 0.89 | 0.79 | 0.018 |
| Family B only | 0.19 | 0.04 | 0.485 |
| Family C only | 0.87 | 0.76 | 0.128 |

[1] "Family" as a control variable for all subjects

Table S2 Correlation of the number of haplotypes per 100 single cells with age

| Samples | r | $r^2$ | *P*-value |
|---|---|---|---|
| All 51 samples from 49 subjects | 0.44 | 0.20 | 0.001 |
| All samples excluding the three cord blood samples and one adolescent sample | 0.42 | 0.17 | 0.003 |
| All samples (controlling for family)[1] | 0.40 | — | 0.005 |
| All samples excluding the three cord blood samples and one adolescent sample (controlling for family)[1] | 0.40 | — | 0.006 |
| Family A only | 0.88 | 0.77 | 0.022 |
| Family B only | 0.16 | 0.03 | 0.554 |
| Family C only | 0.87 | 0.75 | 0.135 |

[1] "Family" as a control variable for all subjects

**Table S3**. Aggregate mtDNA sequence in single $CD34^+$ cells from members in the four families and three cord blood samples

| Family / Donor | Sequence variation | Haplogroup |
|---|---|---|
| Family A | 16168-16192-16256-16266-16294-16526-73-263-455delT-(523-524)insAC | U5a |
| Family B | 16150-16189-16311-152-195-263-270 | H11 |
| Family C / C01 | 16192-16256-16270-16311-16399-73-199-263 | U5a1 |
| Family C / C02 | 16145-16176A-16223-16390-16519-73-152-263 | N1b |
| Family C / C03 | 16126-16294-16296-16304-16519-73-263 | T2b |
| Family D | 16192-16256-16270-16362-16399-16428-73-249-263 | U5a1 |
| CB-1 | 16129-16223-16391-16519-73-152-199-204-207-250-263-573+CCC | I2'3 |
| CB-2 | 16111-16223-16290-16319-16360-16362-97-(106-111)del-146-153-235-263 | A2 |
| CB-3 | 16092-16172-16182C-16183C-16189-16223-16266-16362-73-150-263-489-(523-524)delAC | D5a2a |

Note: These maternally related members in each family had same aggregate sequence. Family C has three matrilines, as represented by C-1, C-2, and C-3, respectively. Sequence variation was scored according to the consensus sequence of the single $CD34^+$ cells. Positions were numbered according to the revised Cambridge Reference Sequence (CRS) [1]. Suffixes C and G indicate transversions and ''ins'' indicates insertions. Insertions were recorded at the last possible site. The length mutations of the C-tract in regions 16184-16193 and 303-309 were not included. All samples had 315insC in the second hypervariable segment of mtDNA control region. Each mtDNA was assigned to respective haplogroup based on the classification system of world mtDNA phylogeny [2].

**Supplementary references**:

[1] Andrews RM, Kubacka I, Chinnery PF, et al. Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. Nat Genet 1999;23:147.

[2] van Oven M, Kayser M. Updated comprehensive phylogenetic tree of global human mitochondrial DNA variation. Hum Mutat 2009;30:E386-394

Table S4. mtDNA sequence variation in single CD34$^+$ cells from healthy donors in four families

| Sample | Mutation | No. | Haplotype | Haplotype defined by substitutions |
|---|---|---|---|---|
| ***Family A*** | | | | |
| A-1 | Aggregate sequence | 39 | 1 | |
| | 16076C>Y, 16174C>Y | 1 | 2 | 1 |
| | 16079C>Y, 523-524insAC/non-ins | 1 | 3 | 2 |
| | 16129G>A, 16310G>R | 1 | 4 | 3 |
| | 16129G>R | 1 | 5 | 4 |
| | 16167C>Y, 16392T>Y, 371C>Y | 1 | 6 | 5 |
| | 16185C>Y, 7C | 1 | 7 | 6 |
| | 16192C>Y, 8C/9C | 1 | 8 | 7 |
| | 16224T>Y | 1 | 9 | 8 |
| | 16234C>Y, 40T>Y, 523-524insAC/non-ins | 1 | 10 | 9 |
| | 16275A>R | 1 | 11 | 10 |
| | 16283A>R | 1 | 12 | 11 |
| | 16286C>T | 1 | 13 | 12 |
| | 16287C>Y, 16336G>R | 1 | 14 | 13 |
| | 16295C>Y, 123A>R | 1 | 15 | 14 |
| | 16299A>R | 1 | 16 | 15 |
| | 16304T>Y, 78C>Y, 8C/9C, 523-524insAC/non-ins | 1 | 17 | 16 |
| | 16308T>Y, 16545T>Y, 91C>Y | 1 | 18 | 17 |
| | 16338A>R | 1 | 19 | 18 |
| | 16381T>Y, 16449C>Y, 523-524ins2AC/insAC | 1 | 20 | 19 |
| | 16445T>Y, 436C>Y | 1 | 21 | 20 |
| | 16447C>A | 1 | 22 | 21 |
| | 16468T>Y, 523-524insAC/non-ins | 1 | 23 | 22 |
| | 16476A>R, 17C>Y | 1 | 24 | 23 |
| | 16484T>Y | 1 | 25 | 24 |
| | 16536C>Y | 1 | 26 | 25 |
| | 72T>Y | 1 | 27 | 26 |
| | 127T>Y | 1 | 28 | 27 |
| | 129T>Y, 8C/9C | 1 | 29 | 28 |
| | 204T>C | 1 | 30 | 29 |
| | 217T>Y | 1 | 31 | 30 |
| | 220T>Y | 1 | 32 | 31 |
| | 233T>Y, 412G>R, 7C/8C | 1 | 33 | 32 |
| | 266T>Y, 8C/9C | 1 | 34 | 33 |
| | 279T>G | 1 | 35 | 34 |
| | 368A>R | 1 | 36 | 35 |
| | 391T>C | 1 | 37 | 36 |
| | 430T>T/A, 437C>Y | 1 | 38 | 37 |
| | 438C>T, 556A>G | 1 | 39 | 38 |
| | 455non-del | 1 | 40 | |
| | 460T>Y, 8C/9C | 1 | 41 | 39 |
| | 469C>Y | 1 | 42 | 40 |
| | 572C>Y, 594C>Y | 1 | 43 | 41 |
| | 579T>Y | 1 | 44 | 42 |
| | 628C>Y | 1 | 45 | 43 |
| | 523-524insAC/non-ins | 1 | 46 | |
| | 523-524non-ins | 1 | 47 | |
| | 7C | 1 | 48 | |
| | 7C/8C | 1 | 49 | |
| | 8C/9C | 4 | 50 | |
| A-2 | Aggregate sequence | 52 | 1 | |
| | 16017T>Y, 16132A>R | 1 | 2 | 1 |
| | 16064T>Y | 1 | 3 | 2 |

| | | | | |
|---|---|---|---|---|
| | 16075T>Y | 1 | 4 | 3 |
| | 16121T>Y | 1 | 5 | 4 |
| | 16153G>R | 1 | 6 | 5 |
| | 16159C>Y, 99T>Y, 7C/8C | 1 | 7 | 6 |
| | 16172T>Y | 1 | 8 | 7 |
| | 16175A>R, 71G>A, 8C/9C | 1 | 9 | 8 |
| | 16198T>Y, 16234C>Y, 214A>G | 1 | 10 | 9 |
| | 16209T>Y, 141C>Y, 495C>T | 1 | 11 | 10 |
| | 16298T>Y | 1 | 12 | 11 |
| | 16356T>Y | 1 | 13 | 12 |
| | 16378C>Y, 387C>Y, 523-524insAC/non-ins | 1 | 14 | 13 |
| | 16549C>Y, 523-524insAC/non-ins | 1 | 15 | 14 |
| | 16559A>R, 292T>C | 1 | 16 | 15 |
| | 109G>R, 8C/9C | 1 | 17 | 16 |
| | 172T>Y, 225G>R, 7C/8C | 1 | 18 | 17 |
| | 187G>G/C | 1 | 19 | 18 |
| | 197A>R | 1 | 20 | 19 |
| | 215A>G, 8C/9C | 1 | 21 | 20 |
| | 290-291delAA/non-del | 1 | 22 | |
| | 305C>Y, 7C | 1 | 23 | 21 |
| | 323G>R | 1 | 24 | 22 |
| | 333T>Y | 1 | 25 | 23 |
| | 393T>Y | 1 | 26 | 24 |
| | 426A>A/T | 1 | 27 | 25 |
| | 488A>G | 1 | 28 | 26 |
| | 574A>R | 1 | 29 | 27 |
| | 578T>Y | 1 | 30 | 28 |
| | 523-524insAC/non-ins | 2 | 31 | |
| | 7C | 2 | 32 | |
| | 7C/8C | 3 | 33 | |
| | 7C/8C/9C | 1 | 34 | |
| | 8C/9C | 3 | 35 | |
| A-3 | Aggregate sequence | 55 | 1 | |
| | 16034G>R | 1 | 2 | 1 |
| | 16051A>R | 1 | 3 | 2 |
| | 16052C>Y, 16263T>Y | 1 | 4 | 3 |
| | 16083C>Y, 7C/8C/9C | 1 | 5 | 4 |
| | 16189T>C, 16191insC/non-ins, 7C | 1 | 6 | 5 |
| | 16189T>C, 7C | 1 | 7 | |
| | 16189T>Y, 7C | 1 | 8 | 6 |
| | 16192C>Y, 112C>Y | 1 | 9 | 7 |
| | 16192C>Y, 8C/9C | 1 | 10 | 8 |
| | 16221C>Y | 1 | 11 | 9 |
| | 16267C>Y, 80C>Y | 1 | 12 | 10 |
| | 16311T>C | 1 | 13 | 11 |
| | 16324T>Y, 204T>C, 7C | 1 | 14 | 12 |
| | 16380C>Y | 1 | 15 | 13 |
| | 16468T>Y | 1 | 16 | 14 |
| | 16568T>Y, 523-524insAC/non-ins | 1 | 17 | 15 |
| | 12T>Y, 523-524insAC/non-ins | 1 | 18 | 16 |
| | 310T>Y (C-tract not counted) | 2 | 19 | 17 |
| | 385A>R, 8C/9C | 1 | 20 | 18 |
| | 512A>R | 1 | 21 | 19 |
| | 585A>R, 8C/9C | 1 | 22 | 20 |
| | 7C/8C | 5 | 23 | |
| | 7C/8C/9C | 3 | 24 | |
| | 8C/9C | 8 | 25 | |

| | | | | |
|---|---|---|---|---|
| A-4 | Aggregate sequence | 57 | 1 | |
| | 16129G>R, 466T>C | 1 | 2 | 1 |
| | 16250C>Y | 1 | 3 | 2 |
| | 16267C>Y | 1 | 4 | 3 |
| | 16544T>Y, 523-524non-ins | 1 | 5 | 4 |
| | 215A>G | 2 | 6 | 5 |
| | 215A>G, 523-524insAC/non-ins | 1 | 7 | |
| | 217T>Y, 523-524insAC/non-ins | 1 | 8 | 6 |
| | 227A>R | 1 | 9 | 7 |
| | 279T>G/T | 1 | 10 | 8 |
| | 523-524insAC/non-ins | 10 | 11 | |
| | 7/8C | 3 | 12 | |
| | 8/9C | 13 | 13 | |
| A-5 | Aggregate sequence | 53 | 1 | |
| | 16172T>Y | 1 | 2 | 1 |
| | 16192C (same to CRS), 189A>G | 1 | 3 | 2 |
| | 16197C>Y, 8/9C | 1 | 4 | 3 |
| | 16249T>Y, 523-524insAC/non-ins | 1 | 5 | 4 |
| | 16251C>Y, 523-524insAC/non-ins | 1 | 6 | 5 |
| | 16272A>R | 1 | 7 | 6 |
| | 16309A>R, 523-524non-ins | 1 | 8 | 7 |
| | 16325T>Y | 1 | 9 | 8 |
| | 16452T>Y, 8/9C | 1 | 10 | 9 |
| | 16482insA/non-ins, 204T>Y | 1 | 11 | 10 |
| | 10T>Y | 1 | 12 | 11 |
| | 18C>Y | 1 | 13 | 12 |
| | 146T>Y | 1 | 14 | 13 |
| | 200A>R, 8/9C | 1 | 15 | 14 |
| | 265T>Y, 266T>Y | 1 | 16 | 15 |
| | 298C>Y | 1 | 17 | 16 |
| | 310T>Y | 1 | 18 | 17 |
| | 346T>Y | 1 | 19 | 18 |
| | 401T>Y | 1 | 20 | 19 |
| | 472A>R | 1 | 21 | 20 |
| | 627G>R, 8C/9C | 1 | 22 | 21 |
| | 7/8C | 1 | 23 | |
| | 7/8C/9C | 2 | 24 | |
| | 8/9C | 10 | 25 | |
| | 523-524insAC/ins2AC | 1 | 26 | |
| | 523-524insAC/non-ins | 5 | 27 | |
| A-6 | Aggregate sequence | 63 | 1 | |
| | 16079C>Y, 1G>R | 1 | 2 | 1 |
| | 16145G>A | 2 | 3 | 2 |
| | 16145G>R | 8 | 4 | 3 |
| | 16145G>R, 8C/9C | 1 | 5 | |
| | 16263delT/non-del | 1 | 6 | |
| | 150C>Y | 1 | 7 | 4 |
| | 585A>R | 1 | 8 | 5 |
| | 7C/8C | 1 | 9 | |
| | 8C/9C | 10 | 10 | |
| | 8C/9C, 523-524insAC/non-ins | 1 | 11 | |
| | 523-524insAC/non-ins | 5 | 12 | |
| | | 95 | | |
| ***Family B*** | | | | |
| B-1 | Aggregate sequence (9C/10C/11C/12C) | 21 | 1 | |
| | 16049G>R, 182C>Y, 9C/10C/11C | 1 | 2 | 1 |
| | 16093T>Y, 9C/10C/11C | 1 | 3 | 2 |

| | | | | |
|---|---|---|---|---|
| | 16110G>R, 9C/10C/11C | 1 | 4 | 3 |
| | 16129G>A, 16540C>Y, 9C/10C | 1 | 5 | 4 |
| | 16131T>C, 9C/10C | 1 | 6 | 5 |
| | 16187C>Y, 9C/10C/11C/12C | 1 | 7 | 6 |
| | 16264C>Y, 570C>Y, 9C/10C | 1 | 8 | 7 |
| | 16295C>Y, 9C/10C/11C | 1 | 9 | 8 |
| | 16319G>R, 8C/9C/10C | 1 | 10 | 9 |
| | 16321C>Y, 9C/10C/11C/12C | 1 | 11 | 10 |
| | 16400C>Y, 9C/10C/11C | 1 | 12 | 11 |
| | 16408C>Y, 540A>R, 8C/9C/10C | 1 | 13 | 12 |
| | 16418T>Y, 466T>Y, 9C/10C/11C | 1 | 14 | 13 |
| | 16475T>Y, 9C/10C | 1 | 15 | 14 |
| | 16529T>Y, 9C/10C | 1 | 16 | 15 |
| | 16544T>Y, 9C/10C/11C/12C | 1 | 17 | 16 |
| | 14T>Y, 9C/10C/11C | 1 | 18 | 17 |
| | 48C>Y, 9C/10C | 1 | 19 | 18 |
| | 64C>T, 9C/10C/11C/12C | 1 | 20 | 19 |
| | 112C>Y, 9C/10C/11C | 1 | 21 | 20 |
| | 113C>Y, 9C/10C/11C | 1 | 22 | 21 |
| | 133T>Y, 204T>C, 9C/10C/11C | 1 | 23 | 22 |
| | 181A>R, 9C/10C/11C/12C | 1 | 24 | 23 |
| | 182C>Y, 8C/9C/10C | 1 | 25 | 24 |
| | 182C>Y, 9C/10C/11C/12C | 1 | 26 | |
| | 230A>R, 9C/10C/11C/12C | 1 | 27 | 25 |
| | 305C>Y, 9C/10C/11C | 1 | 28 | 26 |
| | 368A>R, 492A>R, 9C/10C/11C | 1 | 29 | 27 |
| | 436C>Y, 9C/10C/11C | 1 | 30 | 28 |
| | 470A>R, 9C/10C | 1 | 31 | 29 |
| | 490A>R, 9C/10C/11C | 1 | 32 | 30 |
| | 509C>Y, 9C/10C/11C | 1 | 33 | 31 |
| | 630C>Y, 9C/10C/11C/12C | 1 | 34 | 32 |
| | 8C/9C | 1 | 35 | |
| | 8C/9C/10C | 2 | 36 | |
| | 8C/9C/10C/11C | 3 | 37 | |
| | 9C/10C | 15 | 38 | |
| | 9C/10C/11C | 20 | 39 | |
| B-2 | Aggregate sequence (9C/10C) | 21 | 1 | |
| | 16024T>Y, 16381T>Y, 9C/10C | 1 | 2 | 1 |
| | 16068T>Y, 9C/10C | 1 | 3 | 2 |
| | 16093T>Y, 9C/10C/11C | 1 | 4 | 3 |
| | 16100A>R, 9C/10C | 1 | 5 | 4 |
| | 16131T>Y, 9C/10C/11C/12C | 1 | 6 | 5 |
| | 16147C>Y, 9C/10C/11C/12C | 1 | 7 | 6 |
| | 16150C>Y, 291delA/non-del, 9C/10C/11C | 1 | 8 | 7 |
| | 16187C>Y, 9C/10C/11C/12C | 1 | 9 | 8 |
| | 16256C>Y, 9C/10C/11C/12C | 1 | 10 | 9 |
| | 16258A>R, 9C/10C/11C/12C | 1 | 11 | 10 |
| | 16274G>R, 405T>C, 9C/10C/11C | 1 | 12 | 11 |
| | 16278C>T, 9C/10C | 1 | 13 | 12 |
| | 16298T>Y, 422T>Y, 9C/10C/11C | 1 | 14 | 13 |
| | 16318A>R, 9C/10C/11C/12C | 1 | 15 | 14 |
| | 16356T>Y, 296C>Y, 9C/10C/11C | 1 | 16 | 15 |
| | 16388G>R, 16466A>R, 521A>R, 9C/10C/11C/12C | 1 | 17 | 16 |
| | 16430A>R, 9C/10C/11C | 1 | 18 | 17 |
| | 16438G>R, 9C/10C/11C/12C | 1 | 19 | 18 |
| | 16440T>Y, 8C/9C | 1 | 20 | 19 |
| | 16443T>Y, 9C/10C/11C/12C | 1 | 21 | 20 |
| | 16473G>R, 8C/9C/10C | 1 | 22 | 21 |

| | | | | |
|---|---|---|---|---|
| | 67G>R, 9C/10C/11C/12C | 1 | 23 | 22 |
| | 69G>R, 9C/10C | 1 | 24 | 23 |
| | 105C>Y, 298C>Y, 9C/10C/11C | 1 | 25 | 24 |
| | 111A>R, 9C/10C/11C/12C | 1 | 26 | 25 |
| | 151C>Y, 581A>R, 8C/9C/10C/11C | 1 | 27 | 26 |
| | 204T>Y, 9C/10C/11C | 1 | 28 | 27 |
| | 371C>Y, 9C/10C/11C/12C | 1 | 29 | 28 |
| | 392T>Y, 9C/10C | 1 | 30 | 29 |
| | 419A>R, 9C/10C | 1 | 31 | 30 |
| | 448A>R, 9C/10C/11C | 1 | 32 | 31 |
| | 453T>Y, 9C/10C/11C/12C | 1 | 33 | 32 |
| | 483C>Y, 9C/10C | 1 | 34 | 33 |
| | 515A>R, 9C/10C | 1 | 35 | 34 |
| | 520C>Y, 9C/10C/11C/12C | 1 | 36 | 35 |
| | 573delC/non-del, 9C/10C | 1 | 37 | |
| | 595C>Y, 9C/10C/11C | 1 | 38 | 36 |
| | 8C/9C/10C | 2 | 39 | |
| | 8C/9C/10C/11C | 3 | 40 | |
| | 9C/10C/11C | 18 | 41 | |
| | 9C/10C/11C/12C | 13 | 42 | |
| B-3 | Aggregate sequence (9C/10C/11C) | 15 | 1 | |
| | 16014C>Y, 272A>R, 9C/10C/11C | 1 | 2 | 1 |
| | 16024T>Y, 16086T>Y, 9C/10C/11C | 1 | 3 | 2 |
| | 16032T>Y, 308C>Y, 9C/10C | 1 | 4 | 3 |
| | 16062A>A/T, 9C/10C/11C | 1 | 5 | 4 |
| | 16073C>Y, 9C/10C/11C/12C | 1 | 6 | 5 |
| | 16081A>R, 9C/10C/11C/12C | 1 | 7 | 6 |
| | 16093T>C, 9C/10C/11C/12C | 1 | 8 | 7 |
| | 16105T>Y, 16396T>Y, 9C/10C/11C/12C | 1 | 9 | 8 |
| | 16110G>R, 9C/10C/11C | 1 | 10 | 9 |
| | 16129G>R, 152T>Y, 9C/10C | 1 | 11 | 10 |
| | 16129G>R, 9C/10C/11C/12C | 1 | 12 | 11 |
| | 16131T>C, 146T>Y, 9C/10C/11C/12C | 1 | 13 | 12 |
| | 16131T>C, 8C/9C | 1 | 14 | 13 |
| | 16181A>C, 9C/10C/11C/12C | 1 | 15 | 14 |
| | 16184C>Y, 183A>R, 9C/10C/11C/12C | 1 | 16 | 15 |
| | 16187C>Y, 198C>Y, 9C/10C/11C | 1 | 17 | 16 |
| | 16218C>Y, 523-524delAC/non-del, 9C/10C/11C | 1 | 18 | 17 |
| | 16234C>Y, 9C/10C | 1 | 19 | 18 |
| | 16264C>Y, 9C/10C/11C/12C | 1 | 20 | 19 |
| | 16336G>R, 16563C>Y, 71G>R, 9C/10C/11C/12C | 1 | 21 | 20 |
| | 16362T>Y, 9C/10C/11C | 1 | 22 | 21 |
| | 16488C>T, 10T>Y, 111A>G, 10C/11C/12C | 1 | 23 | 22 |
| | 16513T>Y, 9C/10C | 1 | 24 | 23 |
| | 7A>R, 9C/10C/11C/12C | 1 | 25 | 24 |
| | 22T>Y, 178A>R, 573insC, 8C/9C/10C | 1 | 26 | 25 |
| | 110C>Y, 9C/10C/11C/12C | 1 | 27 | 26 |
| | 125T>T/A, 8C/9C/10C/11C | 1 | 28 | 27 |
| | 133T>Y, 453T>Y, 9C/10C | 1 | 29 | 28 |
| | 146T>Y, 9C/10C/11C/12C | 1 | 30 | 29 |
| | 153A>A/C, 9C/10C | 1 | 31 | 30 |
| | 157T>Y, 9C/10C/11C/12C | 1 | 32 | 31 |
| | 169A>R, 9C/10C/11C | 1 | 33 | 32 |
| | 214A>R, 9C/10C/11C | 1 | 34 | 33 |
| | 304C>A, 11C/12C | 1 | 35 | 34 |
| | 304C>Y, 544C>Y, 8C/9C/10C/11C | 1 | 36 | 35 |
| | 307C>Y, 9C/10C | 1 | 37 | 36 |
| | 310T>Y, 540A>R, 9C/10C | 1 | 38 | 37 |

| | | | | |
|---|---|---|---|---|
| | 370C>Y, 9C/10C/11C/12C | 1 | 39 | 38 |
| | 439A>A/T, 9C/10C | 1 | 40 | 39 |
| | 454T>Y, 9C/10C/11C | 1 | 41 | 40 |
| | 489T>C, 8C/9C/10C/11C | 1 | 42 | 41 |
| | 573insC/non-ins, 9C/10C | 1 | 43 | |
| | 609C>Y, 9C/10C/11C | 1 | 44 | 42 |
| | 8C/9C | 4 | 45 | |
| | 8C/9C/10C | 1 | 46 | |
| | 9C/10C | 9 | 47 | |
| | 9C/10C/11C/12C | 14 | 48 | |
| B-4 | Aggregate sequence (9C/10C/11C) | 22 | 1 | |
| | 16026C>T, 9C/10C/11C | 1 | 2 | 1 |
| | 16086T>C, 454T>Y, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16099C>Y, 8C/9C/10C | 1 | 4 | 3 |
| | 16115C>Y, 9C/10C/11C | 1 | 5 | 4 |
| | 16150C>Y, 511C>Y, 8C/9C/10C/11C | 1 | 6 | 5 |
| | 16179C>T, 9C/10C/11C | 1 | 7 | 6 |
| | 16181A>C, 9C/10C/11C | 1 | 8 | 7 |
| | (16194-16195)AT>CC, 9C/10C/11C | 1 | 9 | 8 |
| | 16214C>T, 9C/10C/11C/12C | 1 | 10 | 9 |
| | 16256C>Y, 8C/9C/10C/11C | 1 | 11 | 10 |
| | 16321C>Y, 9C/10C/11C/12C | 1 | 12 | 11 |
| | 16368T>C, 8C/9C/10C | 1 | 13 | 12 |
| | 16381T>Y, 9C/10C/11C/12C | 1 | 14 | 13 |
| | 16387A>R, 9C/10C/11C | 1 | 15 | 14 |
| | 16542C>Y, 9C/10C/11C | 1 | 16 | 15 |
| | 7A>R, 424T>Y, 9C/10C | 1 | 17 | 16 |
| | 43C>Y, 9C/10C/11C | 1 | 18 | 17 |
| | 107G>R, 210A>R, 9C/10C/11C/12C | 1 | 19 | 18 |
| | 161T>Y, 9C/10C/11C/12C | 1 | 20 | 19 |
| | 162C>Y, 9C/10C/11C | 1 | 21 | 20 |
| | 178A>R, 8C/9C/10C/11C | 1 | 22 | 21 |
| | 195T>Y, 9C/10C/11C | 1 | 23 | 22 |
| | 198C>Y, 9C/10C/11C/12C | 1 | 24 | 23 |
| | 204T>C, 9C/10C/11C | 1 | 25 | 24 |
| | 421T>Y, 9C/10C | 1 | 26 | 25 |
| | 508A>R, 8C/9C/10C/11C | 1 | 27 | 26 |
| | 514C>Y, 9C/10C/11C/12C | 1 | 28 | 27 |
| | 593T>C, 8C/9C/10C | 1 | 29 | 28 |
| | 595C>Y, 9C/10C/11C/12C | 1 | 30 | 29 |
| | 523-524delAC/non-del, 9C/10C/11C | 1 | 31 | |
| | 8C/9C/10C | 4 | 32 | |
| | 8C/9C/10C/11C | 2 | 33 | |
| | 9C/10C | 18 | 34 | |
| | 9C/10C/11C/12C | 20 | 35 | |
| B-5 | Aggregate sequence (9C/10C/11C/12C) | 25 | 1 | |
| | 16042G>A, 2A>R, 463C>Y, 8C/9C/10C/11C | 1 | 2 | 1 |
| | 16069C>Y, 16129G>R, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16087A>R, 16268C>Y, 16283A>R, 8C/9C | 1 | 4 | 3 |
| | 16093T>C, 9C/10C/11C | 1 | 5 | 4 |
| | 16129G>A, 9C/10C/11C | 1 | 6 | 5 |
| | 16129G>R, 9C/10C/11C/12C | 1 | 7 | 6 |
| | 16140T>Y, 9C/10C/11C/12C | 1 | 8 | 7 |
| | 16190C>Y, 9C/10C | 1 | 9 | 8 |
| | 16214C>Y, 16463A>R, 149T>Y, 9C/10C/11C/12C/13C | 1 | 10 | 9 |
| | 16261C>Y, 16559A>R, 8C/9C | 1 | 11 | 10 |
| | 16262C>Y, 9C/10C/11C | 1 | 12 | 11 |

| | | | | |
|---|---|---|---|---|
| | 16269A>R, 146T>C, 9C/10C/11C/12C | 1 | 13 | 12 |
| | 16282C>Y, 9C/10C | 1 | 14 | 13 |
| | 16291C>Y, 9C/10C/11C/12C | 1 | 15 | 14 |
| | 16328C>Y, 16526G>R, 9C/10C | 1 | 16 | 15 |
| | 16353C>Y, 16548C>Y, 242C>Y, 9C/10C/11C/12C | 1 | 17 | 16 |
| | 16374A>R, 9C/10C | 1 | 18 | 17 |
| | 16411C>Y, 9C/10C/11C/12C | 1 | 19 | 18 |
| | 2A>R, 71delG/non-del, 9C/10C/11C/12C, 523-524delAC/non-del | 1 | 20 | 19 |
| | 21A>R, 9C/10C/11C | 1 | 21 | 20 |
| | 24A>R, 9C/10C | 1 | 22 | 21 |
| | 51T>Y, 9C/10C/11C | 1 | 23 | 22 |
| | 63T>Y, 8C/9C | 1 | 24 | 23 |
| | 97G>A, 9C/10C/11C | 1 | 25 | 24 |
| | 142T>Y, 9C/10C/11C | 1 | 26 | 25 |
| | 143G>R, 9C/10C/11C | 1 | 27 | 26 |
| | 144C>Y, 9C/10C, 523-524delAC/non-del | 1 | 28 | 27 |
| | 146T>C, 220T>Y, 9C/10C/11C/12C | 1 | 29 | 28 |
| | 151C>Y, 9C/10C | 1 | 30 | 29 |
| | 155T>Y, 9C/10C/11C/12C | 1 | 31 | 30 |
| | 204T>C, 9C/10C | 1 | 32 | 31 |
| | 204T>C, 9C/10C/11C | 1 | 33 | |
| | 217T>C, 9C/10C | 1 | 34 | 32 |
| | 246T>Y, 9C/10C/11C | 1 | 35 | 33 |
| | 247G>R, 9C/10C/11C | 1 | 36 | 34 |
| | 435C>Y, 500C>Y, 9C/10C/11C | 1 | 37 | 35 |
| | 436C>Y, 9C/10C | 1 | 38 | 36 |
| | 462C>Y, 9C/10C | 1 | 39 | 37 |
| | 519A>A/T, 9C/10C/11C | 1 | 40 | 38 |
| | 521A>R, 9C/10C/11C | 1 | 41 | 39 |
| | 525C>Y, 9C/10C/11C/12C | 1 | 42 | 40 |
| | 8C/9C | 1 | 43 | |
| | 8C/9C/10C | 1 | 44 | |
| | 8C/9C/10C/11C | 1 | 45 | |
| | 8C/9C/10C/11C/12C | 1 | 46 | |
| | 9C/10C | 12 | 47 | |
| | 9C/10C/11C | 12 | 48 | |
| | 9C/10C/11C/12C/13C | 1 | 49 | |
| B-6 | Aggregate sequence (9C/10C/11C) | 22 | 1 | |
| | 16020T>Y, 195T>Y, 9C/10C/11C/12C | 1 | 2 | 1 |
| | 16029T>Y, 182C>T, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16029T>Y, 577G>R, 9C/10C | 1 | 4 | 3 |
| | 16036G>R, 9C/10C | 1 | 5 | 4 |
| | 16135delA/non-del, 9C/10C | 1 | 6 | |
| | 16150C>Y, 9C/10C/11C/12C | 1 | 7 | 5 |
| | 16151C>Y, 16256C>Y, 9C/10C | 1 | 8 | 6 |
| | 16165A>R, 16374A>R, 9C/10C | 1 | 9 | 7 |
| | 16173C>Y, 9C/10C/11C | 1 | 10 | 8 |
| | 16177A>R, 9C/10C/11C | 1 | 11 | 9 |
| | 16235A>R, 9C/10C | 1 | 12 | 10 |
| | 16249T>Y, 9C/10C/11C | 1 | 13 | 11 |
| | 16278C>Y, 9C/10C/11C/12C | 1 | 14 | 12 |
| | 16293A>R, 9C/10C/11C | 1 | 15 | 13 |
| | 16293A>R, 9C/10C/11C, 523-524delAC/non-del | 1 | 16 | |
| | 16312A>R, 2A>R, 152T>Y, 182C>Y, 9C/10C/11C/12C | 1 | 17 | 14 |
| | 16314A>R, 16423A>R, 9C/10C/11C | 1 | 18 | 15 |
| | 16368T>C, 9C/10C/11C/12C | 1 | 19 | 16 |
| | 16389G>R, 8C/9C/10C | 1 | 20 | 17 |
| | 16459C>Y, 500C>Y, 507T>T/A, 9C/10C/11C | 1 | 21 | 18 |

| | | | | |
|---|---|---|---|---|
| | 16469T>T/G, 9C/10C/11C/12C | 1 | 22 | 19 |
| | 16482delA/non-del, 9C/10C/11C/12C | 1 | 23 | |
| | 13A>R, 9C/10C/11C | 1 | 24 | 20 |
| | 59T>Y, 8C/9C/10C | 1 | 25 | 21 |
| | 70G>R, 9C/10C/11C | 1 | 26 | 22 |
| | 142T>Y, 9C/10C | 1 | 27 | 23 |
| | 146T>Y, 9C/10C/11C/12C | 1 | 28 | 24 |
| | 149T>Y, 9C/10C/11C | 1 | 29 | 25 |
| | 157T>Y, 9C/10C/11C/12C | 1 | 30 | 26 |
| | 182C>T, 9C/10C/11C | 1 | 31 | 27 |
| | 182C>Y, 9C/10C/11C | 2 | 32 | 28 |
| | 182C>Y, 9C/10C/11C | | | |
| | 182C>T, 9C/10C/11C/12C | 1 | 33 | |
| | 198C>Y, 9C/10C/11C | 1 | 34 | 29 |
| | 208T>Y, 9C/10C/11C/12C | 1 | 35 | 30 |
| | 266T>Y, 9C/10C | 1 | 36 | 31 |
| | 303C>Y, 9C/10C | 1 | 37 | 32 |
| | 306C>Y, 9C/10C/11C | 1 | 38 | 33 |
| | 484A>R, 9C/10C/11C | 1 | 39 | 34 |
| | 487A>G, 9C/10C/11C | 1 | 40 | 35 |
| | 509C>Y, 9C/10C/11C | 1 | 41 | 36 |
| | 553C>Y, 9C/10C/11C | 1 | 42 | 37 |
| | 605T>C, 9C/10C/11C | 1 | 43 | 38 |
| | 8C/9C/10C | 5 | 44 | |
| | 9C/10C | 14 | 45 | |
| | 9C/10C/11C/12C | 10 | 46 | |
| | 9C/10C/11C/12C/13C | 1 | 47 | |
| B-7 | Aggregate sequence (9C/10C/11C) | 22 | 1 | |
| | 16034G>R, 16105T>Y, 16430A>R, 16487A>R, 9C/10C | 1 | 2 | 1 |
| | 16037A>R, 9C/10C/11C | 1 | 3 | 2 |
| | 16046T>Y, 9C/10C/11C | 1 | 4 | 3 |
| | 16073C>Y, 127T>Y, 9C/10C | 1 | 5 | 4 |
| | 16103A>R, 9C/10C/11C | 1 | 6 | 5 |
| | 16114C>Y, 8C/9C/10C/11C | 1 | 7 | 6 |
| | 16129G>R, 9C/10C/11C | 1 | 8 | 7 |
| | 16129G>R, 9C/10C/11C/12C | 1 | 9 | 8 |
| | 16175A>R, 16183A>Y, 9C/10C | 1 | 10 | 9 |
| | 16226A>R, 629T>Y, 9C/10C | 1 | 11 | 10 |
| | 16254A>R, 9C/10C | 1 | 12 | 11 |
| | 16286C>Y, 113C>Y, 635C>Y, 9C/10C/11C | 1 | 13 | 12 |
| | 16482A>R, 9C/10C/11C/12C | 1 | 14 | 13 |
| | 16545T>Y, 9C/10C | 1 | 15 | 14 |
| | 18C>Y, 9C/10C/11C/12C | 1 | 16 | 15 |
| | 91C>Y, 9C/10C/11C/12C | 1 | 17 | 16 |
| | 197A>R, 9C/10C/11C/12C | 1 | 18 | 17 |
| | 272A>R, 9C/10C/11C | 1 | 19 | 18 |
| | 302A>A/C, 9C/10C/11C | 1 | 20 | 19 |
| | 302delA, 9C/10C/11C | 1 | 21 | |
| | 345C>Y, 9C/10C/11C/12C | 1 | 22 | 20 |
| | 392T>Y, 9C/10C | 1 | 23 | 21 |
| | 489T>Y, 9C/10C/11C | 1 | 24 | 22 |
| | 494C>Y, 9C/10C/11C | 1 | 25 | 23 |
| | 502C>Y, 9C/10C/11C | 1 | 26 | 24 |
| | 516C>Y, 8C/9C/10C | 1 | 27 | 25 |
| | 522C>Y, 9C/10C/11C | 1 | 28 | 26 |
| | 537C>Y, 8C/9C/10C/11C | 1 | 29 | 27 |
| | 548C>Y, 9C/10C/11C | 1 | 30 | 28 |
| | 635C>Y, 9C/10C/11C | 1 | 31 | 29 |

| | | | | |
|---|---|---|---|---|
| | 8C/9C/10C | 2 | 32 | |
| | 8C/9C/10C/11C | 1 | 33 | |
| | 8C/9C/10C/11C/12C | 1 | 34 | |
| | 8C/9C/10C/11C/12C, 523-524delAC/non-del | 1 | 35 | |
| | 9C/10C | 22 | 36 | |
| | 9C/10C, 523-524delAC/non-del | 1 | 37 | |
| | 9C/10C/11C/12C | 15 | 38 | |
| | | | | |
| B-8 | Aggregate sequence (9C/10C/11C) | 30 | 1 | |
| | 16053C>Y, 560C>Y, 9C/10C/11C | 1 | 2 | 1 |
| | 16073C>Y, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16119A>R, 16455G>R, 633A>R, 9C/10C/11C/12C | 1 | 4 | 3 |
| | 16129G>R, 9C/10C | 1 | 5 | 4 |
| | 16129G>R, 9C/10C/11C | 1 | 6 | |
| | 16131T>Y, 9C/10C/11C | 1 | 7 | 5 |
| | 16171A>R, 16311T>Y, 9C/10C/11C/12C | 1 | 8 | 6 |
| | 16214C>T, 9C/10C/11C | 1 | 9 | 7 |
| | 16234C>Y, 9C/10C/11C | 1 | 10 | 8 |
| | 16250C>A/C, 16374A>R, 9C/10C/11C | 1 | 11 | 9 |
| | 16260C>Y, 9C/10C/11C/12C | 1 | 12 | 10 |
| | 16262C>Y, 40T>Y, 8C/9C | 1 | 13 | 11 |
| | 16269A>R, 9C/10C | 1 | 14 | 12 |
| | 16345A>A/T, 16354C>Y, 9C/10C/11C | 1 | 15 | 13 |
| | 16367A>R, 146T>Y, 9C/10C/11C/12C | 1 | 16 | 14 |
| | 16370G>R, 9C/10C/11C | 1 | 17 | 15 |
| | 16469T>Y, 9C/10C/11C | 1 | 18 | 16 |
| | 16509T>Y, 9C/10C/11C | 1 | 19 | 17 |
| | 11C>Y, 9C/10C/11C | 1 | 20 | 18 |
| | 22T>Y, 9C/10C/11C | 1 | 21 | 19 |
| | 89T>Y, 542C>Y, 9C/10C/11C/12C | 1 | 22 | 20 |
| | 102A>R, 9C/10C/11C/12C | 1 | 23 | 21 |
| | 146T>Y, 9C/10C/11C/12C | 1 | 24 | 22 |
| | 156A>R, 9C/10C | 1 | 25 | 23 |
| | 178A>R, 9C/10C/11C | 1 | 26 | 24 |
| | 182C>T, 9C/10C/11C/12C | 1 | 27 | 25 |
| | 422T>Y, 8C/9C | 1 | 28 | 26 |
| | 483C>Y, 9C/10C | 1 | 29 | 27 |
| | 540A>R, 9C/10C | 1 | 30 | 28 |
| | 8C/9C | 3 | 31 | |
| | 8C/9C/10C | 1 | 32 | |
| | 9C/10C | 23 | 33 | |
| | 9C/10C/11C, 523-524delAC/non-del | 1 | 34 | |
| | 9C/10C/11C/12C | 7 | 35 | |
| | | | | |
| B-9 | Aggregate sequence (9C/10C/11C/12C) | 20 | 1 | |
| | 16052C>Y, 9C/10C/11C/12C | 1 | 2 | 1 |
| | 16057C>Y, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16084G>R, 16322A>R, 529G>A, 10C/11C/12C | 1 | 4 | 3 |
| | 16088T>Y, 483C>Y, 611G>R, 9C/10C/11C/12C | 1 | 5 | 4 |
| | 16096G>R, 16134C>Y, 9C/10C/11C/12C | 1 | 6 | 5 |
| | 16140T>Y, 9C/10C/11C/12C | 1 | 7 | 6 |
| | 16146A>R, 9C/10C/11C/12C | 1 | 8 | 7 |
| | 16167C>Y, 9C/10C | 1 | 9 | 8 |
| | 16169C>Y, 9C/10C/11C/12C | 1 | 10 | 9 |
| | 16230A>R, 9C/10C | 1 | 11 | 10 |
| | 16239C>Y, 9C/10C | 1 | 12 | 11 |
| | 16265A>R, 9C/10C | 1 | 13 | 12 |
| | 16290C>Y, 193A>R, 9C/10C/11C/12C | 1 | 14 | 13 |
| | 16291C>Y, 9C/10C/11C | 1 | 15 | 14 |

| | | | | |
|---|---|---|---|---|
| | 16309A>R, 9C/10C/11C | 1 | 16 | 15 |
| | 16328C>Y, 9C/10C/11C | 1 | 17 | 16 |
| | 16333A>R, 619T>Y, 9C/10C/11C | 1 | 18 | 17 |
| | 16337C>Y, 9C/10C/11C/12C | 1 | 19 | 18 |
| | 16399A>R, 16509T>Y, 9C/10C | 1 | 20 | 19 |
| | 16443T>Y, 9C/10C/11C | 1 | 21 | 20 |
| | 16468T>Y, 9C/10C | 1 | 22 | 21 |
| | 16497A>R, 9C/10C/11C/12C | 1 | 23 | 22 |
| | 60T>Y, 9C/10C/11C/12C | 1 | 24 | 23 |
| | 142T>Y, 9C/10C/11C/12C | 1 | 25 | 24 |
| | 146T>Y, 9C/10C/11C/12C | 1 | 26 | 25 |
| | 161T>Y, 9C/10C | 1 | 27 | 26 |
| | 169A>R, 527C>Y, 9C/10C/11C | 1 | 28 | 27 |
| | 173T>Y, 9C/10C/11C | 1 | 29 | 28 |
| | 184G>R, 9C/10C/11C | 1 | 30 | 29 |
| | 198C>Y, 8C/9C/10C | 1 | 31 | 30 |
| | 204T>C, 9C/10C/11C/12C | 1 | 32 | 31 |
| | 208T>T/A, 523-524delAC/non-del, 9C/10C/11C/12C | 1 | 33 | 32 |
| | 219A>R, 9C/10C/11C/12C | 1 | 34 | 33 |
| | 230A>R, 9C/10C/11C | 1 | 35 | 34 |
| | 242+TAA/non-ins, 9C/10C/11C/12C | 1 | 36 | |
| | 248A>R, 9C/10C | 1 | 37 | 35 |
| | 310T>C, long Cs | 1 | 38 | 36 |
| | 390A>R, 446A>R, 523-524delAC/non-del, 9C/10C/11C | 1 | 39 | 37 |
| | 404C>Y, 8C/9C | 1 | 40 | 38 |
| | 419A>R, 9C/10C/11C/12C | 1 | 41 | 39 |
| | 466T>Y, 9C/10C/11C | 1 | 42 | 40 |
| | 497C>Y, 9C/10C/11C | 1 | 43 | 41 |
| | 540A>R, 9C/10C/11C | 1 | 44 | 42 |
| | 612A>R, 10C/11C/12C | 1 | 45 | 43 |
| | 523-524delAC/non-del, 8C/9C/10C | 1 | 46 | |
| | 523-524delAC/non-del, 9C/10C | 1 | 47 | |
| | 8C/9C/10C | 2 | 48 | |
| | 8C/9C/10C/11C | 2 | 49 | |
| | 9C/10C | 8 | 50 | |
| | 10C/11C/12C | 2 | 51 | |
| | 9C/10C/11C | 14 | 52 | |
| B-10 | Aggregate sequence (9C/10C/11C) | 23 | 1 | |
| | 16056C>Y, 9C/10C/11C/12C | 1 | 2 | 1 |
| | 16086T>Y, 16122A>R, 8C/9C/10C | 1 | 3 | 2 |
| | 16093T>C, 9C/10C/11C/12C | 1 | 4 | 3 |
| | 16124T>Y, 9C/10C | 1 | 5 | 4 |
| | 16131T>Y, 179T>Y, 9C/10C/11C/12C | 1 | 6 | 5 |
| | 16186C>Y, 9C/10C/11C | 1 | 7 | 6 |
| | 16272A>R, 8C/9C/10C | 1 | 8 | 7 |
| | 16277A>R, 16547C>Y, 8C/9C/10C | 1 | 9 | 8 |
| | 16311T>Y, 8C/9C | 1 | 10 | 9 |
| | 16317A>R, 9C/10C/11C/12C | 1 | 11 | 10 |
| | 16368T>Y, 9C/10C/11C | 1 | 12 | 11 |
| | 16533T>Y, 355C>Y, 9C/10C/11C | 1 | 13 | 12 |
| | 42T>Y, 9C/10C/11C/12C | 1 | 14 | 13 |
| | 56A>R, 523-524delAC/non-del, 9C/10C | 1 | 15 | 14 |
| | 58T>Y, 9C/10C/11C/12C | 1 | 16 | 15 |
| | 142T>Y, 172T>T/G, 9C/10C | 1 | 17 | 16 |
| | 146T>C, 8C/9C | 1 | 18 | 17 |
| | 185G>R, 9C/10C/11C | 1 | 19 | 18 |
| | 190C>Y, 8C/9C/10C | 1 | 20 | 19 |
| | 224T>Y, 8C/9C | 1 | 21 | 20 |

| | | | | |
|---|---|---|---|---|
| | 235A>R, 9C/10C | 1 | 22 | 21 |
| | 238A>R, 250T>Y, 8C/9C/10C/11C | 1 | 23 | 22 |
| | 291A>R, 8C/9C/10C | 1 | 24 | 23 |
| | 403T>Y, 587C>Y, 9C/10C/11C | 1 | 25 | 24 |
| | 405T>Y, 9C/10C | 1 | 26 | 25 |
| | 420C>Y, 564G>R, 8C/9C/10C | 1 | 27 | 26 |
| | 421T>Y, 9C/10C/11C/12C | 1 | 28 | 27 |
| | 497C>Y, 9C/10C/11C/12C | 1 | 29 | 28 |
| | 536C>Y, 9C/10C/11C | 1 | 30 | 29 |
| | 557C>Y, 9C/10C/11C/12C | 1 | 31 | 30 |
| | 523-524insAC/non-ins, 9C/10C/11C | 1 | 32 | |
| | 8C/9C | 5 | 33 | |
| | 8C/9C/10C | 4 | 34 | |
| | 8C/9C/10C/11C | 3 | 35 | |
| | 9C/10C | 7 | 36 | |
| | 9C/10C/11C/12C | 20 | 37 | |
| | | | | |
| B-11 | Aggregate sequence (9C/10C/11C) | 22 | 1 | |
| | 16066A>G, 16123T>Y, 9C/10C/11C | 1 | 2 | 1 |
| | 16103A>R, 421T>Y, 512A>R, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16150C>Y, 9C/10C | 1 | 4 | 3 |
| | 16182A>Y, 9C/10C | 1 | 5 | 4 |
| | 16222C>Y, 9C/10C/11C | 1 | 6 | 5 |
| | 16226A>R, 9C/10C/11C | 1 | 7 | 6 |
| | 16251C>Y, 9C/10C/11C | 1 | 8 | 7 |
| | 16262delC/non-del, 9C/10C/11C | 1 | 9 | |
| | 16291C>Y, 8C/9C/10C/11C | 1 | 10 | 8 |
| | 16368T>Y, 8C/9C | 1 | 11 | 9 |
| | 16386T>Y, 8C/9C | 1 | 12 | 10 |
| | 16411C>Y, 9C/10C/11C | 1 | 13 | 11 |
| | 16425C>Y, 9C/10C | 1 | 14 | 12 |
| | 16442C>Y, 9C/10C/11C | 1 | 15 | 13 |
| | 16459C>Y, 9C/10C/11C | 1 | 16 | 14 |
| | 16490G>R, 9C/10C | 1 | 17 | 15 |
| | 16495C>Y, 9C/10C/11C/12C | 1 | 18 | 16 |
| | 16534A>R, 8C/9C/10C | 1 | 19 | 17 |
| | 16556A>R, 8C/9C | 1 | 20 | 18 |
| | 19C>Y, 9C/10C/11C | 1 | 21 | 19 |
| | 114C>Y, 449T>Y, 9C/10C | 1 | 22 | 20 |
| | 146T>Y, 9C/10C | 1 | 23 | 21 |
| | 174C>Y, 9C/10C/11C | 1 | 24 | 22 |
| | 234A>R, 9C/10C/11C/12C | 1 | 25 | 23 |
| | 304C>Y, 9C/10C | 1 | 26 | 24 |
| | 404C>Y, 9C/10C/11C | 1 | 27 | 25 |
| | 482T>Y, 8C/9C/10C | 1 | 28 | 26 |
| | 538A>R, 9C/10C | 1 | 29 | 27 |
| | 8C/9C | 5 | 30 | |
| | 8C/9C/10C | 5 | 31 | |
| | 8C/9C/10C/11C | 2 | 32 | |
| | 8C/9C/10C/11C/12C | 1 | 33 | |
| | 9C/10C | 20 | 34 | |
| | 9C/10C/11C/12C | 8 | 35 | |
| | | | | |
| B-12 | Aggregate sequence (9C/10C) | 25 | 1 | |
| | 16029T>Y, 153A>R, 8C/9C/10C | 1 | 2 | 1 |
| | 16062A>R, 9C/10C/11C | 1 | 3 | 2 |
| | 16086T>Y, 16129G>A, 8C/9C | 1 | 4 | 3 |
| | 16095C>Y, 9C/10C | 1 | 5 | 4 |
| | 16114C>Y, 8C/9C | 1 | 6 | 5 |

| | | | |
|---|---|---|---|
| 16124T>C, 9C/10C/11C/12C | 1 | 7 | 6 |
| 16129G>A, 16278C>Y, 8C/9C/10C | 1 | 8 | 7 |
| 16129G>A, 16476A>R, 9C/10C | 1 | 9 | 8 |
| 16129G>A, 204T>C, 9C/10C | 1 | 10 | 9 |
| 16129G>A, 462C>Y, 8C/9C/10C | 1 | 11 | 10 |
| 16129G>A, 8C/9C | 5 | 12 | 11 |
| 16129G>A, 8C/9C/10C | 2 | 13 | |
| 16129G>A, 8C/9C/10C/11C | 1 | 14 | |
| 16129G>A, 9C/10C | 6 | 15 | |
| 16129G>A, 9C/10C/11C | 2 | 16 | |
| 16129G>R, 8C/9C | 5 | 17 | 12 |
| 16129G>R, 8C/9C/10C | 1 | 18 | |
| 16129G>R, 8C/9C/10C/11C | 1 | 19 | |
| 16129G>R, 9C/10C | 4 | 20 | |
| 16129G>R, 9C/10C/11C | 3 | 21 | |
| 16129G>R, 9C/10C/11C/12C | 1 | 22 | |
| 16131T>Y, 8C/9C | 1 | 23 | 13 |
| 16136T>T/A, 9C/10C | 1 | 24 | 14 |
| 16137A>R, 9C/10C/11C/12C | 1 | 25 | 15 |
| 16152T>Y, 9C/10C/11C | 1 | 26 | 16 |
| 16158A>R, 16272A>R, 8C/9C/10C | 1 | 27 | 17 |
| 16167C>Y, 9C/10C/11C/12C | 1 | 28 | 18 |
| 16184C>Y, 8C/9C | 1 | 29 | 19 |
| 16187C>Y, 8C/9C | 1 | 30 | 20 |
| 16203A>R, 9C/10C/11C | 1 | 31 | 21 |
| 16211C>Y, 8C/9C/10C | 1 | 32 | 22 |
| 16228C>T, 9C/10C | 1 | 33 | 23 |
| 16250C>Y, 71delG/non-del, 8C/9C | 1 | 34 | 24 |
| 16263delT/non-del, 4C>Y, 9C/10C/11C | 1 | 35 | 25 |
| 16307A>R, 8C/9C/10C, 523-524delAC/non-del | 1 | 36 | 26 |
| 16320C>Y, 8C/9C | 1 | 37 | 27 |
| 16336G>R, 8C/9C/10C/11C | 1 | 38 | 28 |
| 16377C>Y, 8C/9C/10C | 1 | 39 | 29 |
| 16423A>R, 143G>R, 8C/9C/10C | 1 | 40 | 30 |
| 16427C>Y, 9C/10C | 1 | 41 | 31 |
| 16449C>Y, 9C/10C | 1 | 42 | 32 |
| 16463A>R, 9C/10C | 1 | 43 | 33 |
| 16464A>R, 9C/10C/11C | 1 | 44 | 34 |
| 16486A>R, 8C/9C | 1 | 45 | 35 |
| 16519T>C, 8C/9C/10C | 1 | 46 | 36 |
| 39C>Y, 8C/9C/10C | 1 | 47 | 37 |
| 54G>R, 8C/9C | 1 | 48 | 38 |
| 60T>Y, 8C/9C | 1 | 49 | 39 |
| 70G>R, 9C/10C/11C/12C, 523-524delAC/non-del | 1 | 50 | 40 |
| 93A>R, 9C/10C | 1 | 51 | 41 |
| 112C>Y, 9C/10C/11C | 1 | 52 | 42 |
| 133T>Y, 9C/10C/11C | 1 | 53 | 43 |
| 156A>R, 9C/10C/11C/12C | 1 | 54 | 44 |
| 181A>R, 9C/10C | 1 | 55 | 45 |
| 226T>Y, 9C/10C | 1 | 56 | 46 |
| 246T>Y, 8C/9C/10C | 1 | 57 | 47 |
| 266T>Y, 8C/9C | 1 | 58 | 48 |
| 296C>Y, 9C/10C | 1 | 59 | 49 |
| 307C>Y, 9C/10C | 1 | 60 | 50 |
| 381C>Y, 9C/10C | 1 | 61 | 51 |
| 420C>Y, 9C/10C/11C | 1 | 62 | 52 |
| 434C>Y, 9C/10C | 1 | 63 | 53 |
| 439A>R, 8C/9C/10C | 1 | 64 | 54 |
| 468C>Y, 9C/10C | 1 | 65 | 55 |

| | | | | |
|---|---|---|---|---|
| | 515A>R, 9C/10C | 1 | 66 | 56 |
| | 519A>A/T, 9C/10C/11C/12C | 1 | 67 | 57 |
| | 544C>Y, 9C/10C/11C | 1 | 68 | 58 |
| | 570C>Y, 8C/9C | 1 | 69 | 59 |
| | 605T>Y, 8C/9C | 1 | 70 | 60 |
| | 626G>R, 9C/10C/11C | 1 | 71 | 61 |
| | 8C/9C | 22 | 72 | |
| | 8C/9C/10C | 16 | 73 | |
| | 8C/9C/10C/11C | 6 | 74 | |
| | 9C/10C/11C | 16 | 75 | |
| | 9C/10C/11C/12C | 10 | 76 | |
| | | | | |
| B-13 | Aggregate sequence (9C/10C/11C) | 59 | 1 | |
| | 16022T>Y, 9C/10C | 1 | 2 | 1 |
| | 16036G>R, 470A>R, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16037A>R, 9C/10C | 1 | 4 | 3 |
| | 16052C>Y, 9C/10C/11C | 1 | 5 | 4 |
| | 16095C>Y, 9C/10C/11C | 1 | 6 | 5 |
| | 16111C>Y, 9C/10C | 1 | 7 | 6 |
| | 16129G>A, 9C/10C | 1 | 8 | 7 |
| | 16129G>R, 215A>G, 9C/10C/11C | 1 | 9 | 8 |
| | 16134C>Y, 9C/10C | 1 | 10 | 9 |
| | 16137A>R, 9C/10C/11C | 1 | 11 | 10 |
| | 16142C>Y, 204T>Y, 10C/11C/12C | 1 | 12 | 11 |
| | 16154T>Y, 9C/10C/11C/12C | 1 | 13 | 12 |
| | 16176C>Y, 298C>Y, 9C/10C/11C | 1 | 14 | 13 |
| | 16178T>A, 16264C>Y, 8C/9C | 1 | 15 | 14 |
| | 16187C>Y, 9C/10C/11C/12C | 1 | 16 | 15 |
| | 16231T>Y, 16281A>R, 57T>Y, 9C/10C/11C | 1 | 17 | 16 |
| | 16242C>Y, 9C/10C | 1 | 18 | 17 |
| | 16246A>R, 9C/10C/11C | 1 | 19 | 18 |
| | 16265A>R, 9C/10C/11C/12C | 1 | 20 | 19 |
| | 16266C>Y, 9C/10C/11C | 1 | 21 | 20 |
| | 16281A>R, 9C/10C/11C/12C | 1 | 22 | 21 |
| | 16342T>Y, 9C/10C/11C | 1 | 23 | 22 |
| | 16371A>R, 9C/10C/11C/12C | 1 | 24 | 23 |
| | 16381delT/non-del, 9C/10C | 1 | 25 | |
| | 16449C>Y, 9C/10C/11C/12C | 1 | 26 | 24 |
| | 16487A>R, 230A>R, 8C/9C | 1 | 27 | 25 |
| | 16507C>Y, 9C/10C/11C | 1 | 28 | 26 |
| | 16512T>Y, 9C/10C/11C | 1 | 29 | 27 |
| | 16518G>R, 9C/10C/11C/12C | 1 | 30 | 28 |
| | 16520C>T, 8C/9C | 1 | 31 | 29 |
| | 16533T>Y, 189A>R, 9C/10C | 1 | 32 | 30 |
| | 16534A>R, 9C/10C | 1 | 33 | 31 |
| | 16540C>Y, 114C>Y, 9C/10C/11C | 1 | 34 | 32 |
| | 16544T>Y, 9C/10C | 1 | 35 | 33 |
| | 16559A>R, 9C/10C/11C | 1 | 36 | 34 |
| | 2A>R, 9C/10C/11C | 1 | 37 | 35 |
| | 3T>Y, 9C/10C/11C/12C | 1 | 38 | 36 |
| | 3T>Y, 9C/10C/11C/12C | 1 | 39 | 37 |
| | 10T>Y, 16A>R, 9C/10C/11C/12C | 1 | 40 | 38 |
| | 12T>Y, 9C/10C/11C/12C | 1 | 41 | 39 |
| | 41C>Y, 431C>Y, 9C/10C/11C/12C | 1 | 42 | 40 |
| | 55T>Y, 9C/10C/11C/12C | 1 | 43 | 41 |
| | 88T>Y, 9C/10C/11C | 1 | 44 | 42 |
| | 91C>Y, 305C>Y, 9C/10C/11C/12C | 1 | 45 | 43 |
| | 97G>G/T, 8C/9C/10C | 1 | 46 | 44 |
| | 98C>Y, 9C/10C/11C | 1 | 47 | 45 |

| | | | | |
|---|---|---|---|---|
| | 102A>R, 9C/10C | 1 | 48 | 46 |
| | 143G>R, 9C/10C/11C/12C | 1 | 49 | 47 |
| | 146T>Y, 9C/10C | 1 | 50 | 48 |
| | 162C>Y, 9C/10C/11C/12C | 1 | 51 | 49 |
| | 172T>Y, 9C/10C | 1 | 52 | 50 |
| | 216T>Y, 9C/10C | 1 | 53 | 51 |
| | 227A>R, 9C/10C/11C/12C | 1 | 54 | 52 |
| | 239T>Y, 9C/10C/11C | 1 | 55 | 53 |
| | 240A>R, 9C/10C/11C | 1 | 56 | 54 |
| | 258C>Y, 9C/10C | 1 | 57 | 55 |
| | 265T>Y, 9C/10C | 1 | 58 | 56 |
| | 269C>Y, 8C/9C/10C/11C | 1 | 59 | 57 |
| | 270A>R, 9C/10C/11C/12C | 1 | 60 | 58 |
| | 276A>R, 10C/11C/12C | 1 | 61 | 59 |
| | 278A>R, 439A>R, 9C/10C/11C/12C | 1 | 62 | 60 |
| | 279T>Y, 8C/9C/10C/11C | 1 | 63 | 61 |
| | 308C>Y, 9C/10C | 1 | 64 | 62 |
| | 309C>Y, 9C/10C/11C/12C | 1 | 65 | 63 |
| | 399T>Y, 9C/10C | 1 | 66 | 64 |
| | 405T>Y, 8C/9C/10C/11C/12C | 1 | 67 | 65 |
| | 421T>Y, 506C>Y, 9C/10C/11C/12C | 1 | 68 | 66 |
| | 438C>Y, 10C/11C/12C | 1 | 69 | 67 |
| | 454T>Y, 9C/10C | 1 | 70 | 68 |
| | 466T>Y, 9C/10C/11C | 1 | 71 | 69 |
| | 552C>Y, 9C/10C/11C | 1 | 72 | 70 |
| | 8C/9C | 7 | 73 | |
| | 8C/9C/10C | 5 | 74 | |
| | 9C/10C | 22 | 75 | |
| | 9C/10C/11C/12C | 25 | 76 | |
| | 10C/11C/12C | 2 | 77 | |
| B-14 | Aggregate sequence (8C/9C/10C) | 13 | 1 | |
| | 16034G>R, 9C/10C | 1 | 2 | 1 |
| | 16096G>R, 9C/10C/11C/12C | 1 | 3 | 2 |
| | 16102T>Y, 9C/10C | 1 | 4 | 3 |
| | 16109A>A/T, 146T>C, 8C/9C | 1 | 5 | 4 |
| | 16121T>Y, 9C/10C | 1 | 6 | 5 |
| | 16150C>Y, 16377C>Y, 8C/9C | 1 | 7 | 6 |
| | 16169C>T, 16184C>Y, 416T>Y, 9C/10C/11C | 1 | 8 | 7 |
| | 16229T>Y, 9C/10C | 1 | 9 | 8 |
| | 16267C>Y, 113C>Y, 9C/10C/11C/12C | 1 | 10 | 9 |
| | 16275A>R, 8C/9C/10C/11C | 1 | 11 | 10 |
| | 16290C>Y, 8C/9C | 1 | 12 | 11 |
| | 16312A>R, 9C/10C/11C/12C | 1 | 13 | 12 |
| | 16418T>Y, 9C/10C/11C/12C | 1 | 14 | 13 |
| | 16457G>R, 8C/9C | 1 | 15 | 14 |
| | 16481A>A/T, 386C>Y, 9C/10C | 1 | 16 | 15 |
| | 16486A>R, 9C/10C | 1 | 17 | 16 |
| | 16531A>R, 223T>Y, 9C/10C | 1 | 18 | 17 |
| | 16544T>Y, 9C/10C/11C | 1 | 19 | 18 |
| | 12T>Y, 9C/10C | 1 | 20 | 19 |
| | 15C>T, 9C/10C | 1 | 21 | 20 |
| | 136G>R, 9C/10C | 1 | 22 | 21 |
| | 146T>C, 8C/9C | 1 | 23 | 22 |
| | 146T>Y, 497C>Y, 9C/10C/11C | 1 | 24 | 23 |
| | 146T>Y, 8C/9C | 1 | 25 | 24 |
| | 146T>Y, 8C/9C/10C | 1 | 26 | |
| | 146T>Y, 9C/10C | 1 | 27 | |
| | 173T>Y, 9C/10C | 1 | 28 | 25 |

| | | | | |
|---|---|---|---|---|
| | 200A>R, 8C/9C | 1 | 29 | 26 |
| | 204T>C, 9C/10C/11C/12C | 1 | 30 | 27 |
| | 204T>Y, 8C/9C | 1 | 31 | 28 |
| | 204T>Y, 9C/10C/11C | 1 | 32 | |
| | 252T>Y, 8C/9C | 1 | 33 | 29 |
| | 295C>Y, 606A>R, 9C/10C | 1 | 34 | 30 |
| | 297A>R, 8C/9C/10C | 1 | 35 | 31 |
| | 302A>R, 8C/9C/10C | 1 | 36 | 32 |
| | 302delA, 303C>C/A, 9C/10C/11C | 1 | 37 | 33 |
| | 302delA, 304C>Y, 405T>Y, 9C/10C/11C/12C | 1 | 38 | 34 |
| | 302delA, 9C/10C | 1 | 39 | |
| | 369C>Y, 9C/10C/11C/12C | 1 | 40 | 35 |
| | 431C>Y, 8C/9C | 1 | 41 | 36 |
| | 440delA/non-del, 9C/10C/11C | 1 | 42 | |
| | 453T>Y, 8C/9C | 1 | 43 | 37 |
| | 523-524delAC/non-del, 9C/10C/11C | 1 | 44 | |
| | 534C>Y, 8C/9C/10C/11C | 1 | 45 | 38 |
| | 573insC/non-ins, 9C/10C | 1 | 46 | |
| | 587C>Y, 9C/10C | 1 | 47 | 39 |
| | 606A>R, 8C/9C | 1 | 48 | 40 |
| | 8C/9C | 8 | 49 | |
| | 8C/9C/10C/11C | 3 | 50 | |
| | 8C/9C/11C | 1 | 51 | |
| | 9C/10C | 11 | 52 | |
| | 9C/10C/11C | 10 | 53 | |
| | 9C/10C/11C/12C | 3 | 54 | |
| B-15 | Aggregate sequence (9C/10C/11C) | 20 | 1 | |
| | 16038insA/non-ins, 9C/10C/11C | 1 | 2 | |
| | 16049G>R, 16114C>Y, 9C/10C | 1 | 3 | 1 |
| | 16053C>Y, 8C/9C/10C | 1 | 4 | 2 |
| | 16062A>R, 16347T>Y, 9C/10C | 1 | 5 | 3 |
| | 16068T>Y, 16218C>T, 416T>Y, 8C/9C | 1 | 6 | 4 |
| | 16084G>R, 146T>Y, 8C/9C | 1 | 7 | 5 |
| | 16109A>A/T, 8C/9C/10C | 1 | 8 | 6 |
| | 16144T>Y, 8C/9C/10C | 1 | 9 | 7 |
| | 16151C>Y, 9C/10C | 1 | 10 | 8 |
| | 16162A>G, 9C/10C/11C | 1 | 11 | 9 |
| | 16179C>Y, 9C/10C/11C/12C | 1 | 12 | 10 |
| | 16237A>R, 8C/9C/10C | 1 | 13 | 11 |
| | 16249T>Y, 9C/10C | 1 | 14 | 12 |
| | 16268C>Y, 423T>Y, 9C/10C/11C/12C | 1 | 15 | 13 |
| | 16274G>R, 353C>Y, 9C/10C | 1 | 16 | 14 |
| | 16379C>Y, 9C/10C | 1 | 17 | 15 |
| | 16395C>Y, 9C/10C/11C | 1 | 18 | 16 |
| | 16403C>Y, 9C/10C/11C | 1 | 19 | 17 |
| | 16453C>Y, 8C/9C/10C/11C | 1 | 20 | 18 |
| | 16506T>Y, 59T>Y, 9C/10C/11C/12C | 1 | 21 | 19 |
| | 16529T>Y, 8C/9C | 1 | 22 | 20 |
| | 13A>R, 9C/10C/11C/12C | 1 | 23 | 21 |
| | 33C>Y, 9C/10C | 1 | 24 | 22 |
| | 54G>R, 8C/9C/10C | 1 | 25 | 23 |
| | 68G>R, 9C/10C/11C/12C | 1 | 26 | 24 |
| | 74T>Y, 9C/10C/11C | 1 | 27 | 25 |
| | 121G>R, 146T>C, 9C/10C/11C/12C | 1 | 28 | 26 |
| | 125T>Y, 9C/10C | 1 | 29 | 27 |
| | 199T>Y, 490A>R, 9C/10C/11C | 1 | 30 | 28 |
| | 227A>A/T, 384A>R, 9C/10C/11C/12C | 1 | 31 | 29 |
| | 245T>Y, 9C/10C | 1 | 32 | 30 |

| | | | | |
|---|---|---|---|---|
| | 285C>Y, 8C/9C/10C/11C | 1 | 33 | 31 |
| | 298C>Y, 9C/10C/11C | 1 | 34 | 32 |
| | 302delA, 9C/10C | 1 | 35 | |
| | 356C>Y, 8C/9C | 1 | 36 | 33 |
| | 382C>Y, 8C/9C | 1 | 37 | 34 |
| | 422T>Y, 9C/10C/11C | 1 | 38 | 35 |
| | 435C>Y, 8C/9C | 1 | 39 | 36 |
| | 442T>Y, 607C>Y, 9C/10C/11C/12C | 1 | 40 | 37 |
| | 454T>Y, 8C/9C/10C | 1 | 41 | 38 |
| | 482T>Y, 8C/9C | 1 | 42 | 39 |
| | 605T>Y, 9C/10C | 1 | 43 | 40 |
| | 605T>Y, 8C/9C/10C | 1 | 44 | |
| | 626G>R, 8C/9C/10C | 1 | 45 | 41 |
| | 8C/9C | 4 | 46 | |
| | 8C/9C/10C | 3 | 47 | |
| | 8C/9C/10C/11C | 2 | 48 | |
| | 9C/10C | 18 | 49 | |
| | 9C/10C/11C/12C | 5 | 50 | |
| | | | | |
| B-16 | Aggregate sequence (9C/10C/11C) | 25 | 1 | |
| | 16095C>C/G, 9C/10C/11C | 1 | 2 | 1 |
| | 16095C>Y, 9C/10C/11C | 1 | 3 | 2 |
| | 16158A>R, 9C/10C/11C/12C | 1 | 4 | 3 |
| | 16167C>Y, 16185C>Y, 9C/10C | 1 | 5 | 4 |
| | 16173C>Y, 9C/10C | 1 | 6 | 5 |
| | 16176C>Y, 9C/10C/11C/12C | 1 | 7 | 6 |
| | 16177A>A/T, 8C/9C | 1 | 8 | 7 |
| | 16180A>R, 9C/10C/11C | 1 | 9 | 8 |
| | 16260C>Y, 8C/9C | 1 | 10 | 9 |
| | 16286C>Y, 9C/10C/11C/12C | 1 | 11 | 10 |
| | 16291C>Y, 9C/10C | 1 | 12 | 11 |
| | 16384G>R, 9C/10C | 1 | 13 | 12 |
| | 16478C>Y, 9C/10C/11C | 1 | 14 | 13 |
| | 151C>T, 9C/10C/11C/12 | 1 | 15 | 14 |
| | 167C>Y, 9C/10C | 1 | 16 | 15 |
| | 179T>Y, 9C/10C/11C | 1 | 17 | 16 |
| | 204T>Y, 8C/9C | 1 | 18 | 17 |
| | 252T>Y, 9C/10C/11C | 1 | 19 | 18 |
| | 296C>Y, 9C/10C/11C | 1 | 20 | 19 |
| | 303C>Y, 9C/10C/11C/12C | 1 | 21 | 20 |
| | 398T>Y, 9C/10C/11C | 1 | 22 | 21 |
| | 467C>Y, 9C/10C | 1 | 23 | 22 |
| | 569C>Y, 9C/10C | 1 | 24 | 23 |
| | 523-524delAC/non-del, 9C/10C/11C/12C | 1 | 25 | |
| | 8C | 1 | 26 | |
| | 8C/9C | 3 | 27 | |
| | 8C/9C/10C | 3 | 28 | |
| | 8C/9C/10C/11C | 1 | 29 | |
| | 9C/10C | 18 | 30 | |
| | 9C/10C/11C/12C | 21 | 31 | |
| | | | | |
| ***Family C*** | | | | |
| C-1 | aggregate sequence | 32 | 1 | |
| | 16032T>Y | 1 | 2 | 1 |
| | 16055C>Y, 16541A>R | 1 | 3 | 2 |
| | 16129G>A, 16471G>A | 1 | 4 | 3 |
| | 16131T>C | 1 | 5 | 4 |
| | 16131T>C, 452T>Y | 1 | 6 | 5 |
| | 16137A>R, 305C>Y, 7C/8C | 1 | 7 | 6 |

| | | | | |
|---|---|---|---|---|
| | 16140T>Y, 162C>C/A, 519A>A/T | 1 | 8 | 7 |
| | 16149A>A/T, 152T>C | 1 | 9 | 8 |
| | 16171A>A/T, 204T>C | 1 | 10 | 9 |
| | 16189T>Y, 16252A>R | 1 | 11 | 10 |
| | 16189T>Y, 16559A>R | 1 | 12 | 11 |
| | 16210A>R | 1 | 13 | 12 |
| | 16277A>R | 1 | 14 | 13 |
| | 16292delC/non-del | 1 | 15 | |
| | 16297T>Y | 1 | 16 | 14 |
| | 16319G>A | 1 | 17 | 15 |
| | 16323T>Y | 1 | 18 | 16 |
| | 16346G>R, 278A>R, 8C/9C | 1 | 19 | 17 |
| | 16353C>Y, 8C/9C | 1 | 20 | 18 |
| | 16409T>Y, 385A>G | 1 | 21 | 19 |
| | 16411C>Y | 1 | 22 | 20 |
| | 16413T>T/G, 204T>C | 1 | 23 | 21 |
| | 16448T>Y, 109G>R, 8C/9C | 1 | 24 | 22 |
| | 16463A>R | 1 | 25 | 23 |
| | 16474insG/non-ins | 1 | 26 | |
| | 16521A>R | 1 | 27 | 24 |
| | 16538C>Y | 1 | 28 | 25 |
| | 13A>R | 1 | 29 | 26 |
| | 37A>R | 1 | 30 | 27 |
| | 53G>G/T | 1 | 31 | 28 |
| | 55T>Y | 1 | 32 | 29 |
| | 113C>Y, 8C/9C | 1 | 33 | 30 |
| | 126A>A/T, 394C>Y, 471T>Y | 1 | 34 | 31 |
| | 153A>G | 1 | 35 | 32 |
| | 204T>C | 2 | 36 | 33 |
| | 204T>Y, 291delA | 1 | 37 | 34 |
| | 234A>R | 1 | 38 | 35 |
| | 312C>Y | 1 | 39 | 36 |
| | 398T>Y | 1 | 40 | 37 |
| | 438delC/non-del | 1 | 41 | |
| | 453T>Y | 1 | 42 | 38 |
| | 478A>R | 1 | 43 | 39 |
| | 490A>R | 1 | 44 | 40 |
| | 512A>A/C, 7C/8C | 1 | 45 | 41 |
| | 512A>R | 1 | 46 | 42 |
| | 532A>R | 1 | 47 | 43 |
| | 541C>Y | 1 | 48 | 44 |
| | 558C>Y | 1 | 49 | 45 |
| | 7C/8C | 2 | 50 | |
| | 8C/9C | 10 | 51 | |
| C-2 | Aggregate sequence | 29 | 1 | |
| | 16075T>Y | 1 | 2 | 1 |
| | 16092T>Y | 1 | 3 | 2 |
| | 16129G>A | 1 | 4 | 3 |
| | 16151C>Y, 16324T>T/A | 1 | 5 | 4 |
| | 16166A>C | 1 | 6 | 5 |
| | 16213G>A, 5A>R, 71delG | 1 | 7 | 6 |
| | 16219A>R | 1 | 8 | 7 |
| | 16221C>Y | 1 | 9 | 8 |
| | 16251C>Y | 1 | 10 | 9 |
| | 16291C>Y, 16435A>R, 462C>Y | 1 | 11 | 10 |
| | 16302A>R, 617G>R | 1 | 12 | 11 |
| | 16311T>Y, 71delG, 523-524delAC/non-del | 1 | 13 | 12 |
| | 16333A>R | 1 | 14 | 13 |

| | | | | |
|---|---|---|---|---|
| | 16365C>Y, 71delG | 1 | 15 | 14 |
| | 16369G>A, 146T>C | 1 | 16 | 15 |
| | 16377C>T, 71delG | 1 | 17 | 16 |
| | 16406T>Y | 1 | 18 | 17 |
| | 16425C>Y | 1 | 19 | 18 |
| | 16439C>Y | 1 | 20 | 19 |
| | 16476A>R, 72T>Y | 1 | 21 | 20 |
| | 16479T>Y | 1 | 22 | 21 |
| | 16489T>Y, 71delG | 1 | 23 | 22 |
| | 16540C>T | 1 | 24 | 23 |
| | 16563C>Y | 1 | 25 | 24 |
| | 59T>T/G, 71delG | 1 | 26 | 25 |
| | 71delG | 15 | 27 | |
| | 71delG, (311-315)6C/7C | 1 | 28 | |
| | 71delG, 131T>Y, 497C>Y | 1 | 29 | 26 |
| | 71delG, 220T>Y | 1 | 30 | 27 |
| | 71delG, 452T>Y | 1 | 31 | 28 |
| | 71delG, 8C/9C | 1 | 32 | |
| | 71delG/non-del | 2 | 33 | |
| | 72T>Y, 193A>R | 1 | 34 | 29 |
| | 113C>Y | 1 | 35 | 30 |
| | 206T>Y | 1 | 36 | 31 |
| | 242C>Y | 1 | 37 | 32 |
| | 247G>A | 1 | 38 | 33 |
| | 373A>T, 8C/9C | 1 | 39 | 34 |
| | 393T>Y, 414T>G, 7C/8C | 1 | 40 | 35 |
| | 612A>R, 8C/9C | 1 | 41 | 36 |
| | 627G>R | 1 | 42 | 37 |
| | 523-524delAC/non-del | 2 | 43 | |
| | 7C/8C/9C | 3 | 44 | |
| | 8C/9C | 4 | 45 | |
| C-3 | aggregate sequence | 77 | 1 | |
| | 16129G>R | 1 | 2 | 1 |
| | 16221C>Y | 1 | 3 | 2 |
| | 16265A>C | 1 | 4 | 3 |
| | 16266C>Y | 1 | 5 | 4 |
| | 16287C>Y | 1 | 6 | 5 |
| | 16304T (same as CRS) | 1 | 7 | 6 |
| | 146T>Y | 1 | 8 | 7 |
| | 150C>T, 7C/8C | 1 | 9 | 8 |
| | 152T>Y | 2 | 10 | 9 |
| | 189A>R | 2 | 11 | 10 |
| | 215A>R | 1 | 12 | 11 |
| | 222C>Y | 1 | 13 | 12 |
| | 491C>Y | 1 | 14 | 13 |
| | 522C>Y | 1 | 15 | 14 |
| | 593T>Y | 1 | 16 | 15 |
| | 523-524delAC/non-del | 2 | 17 | |
| C-4 | aggregate sequence | 40 | 1 | |
| | 16023G>R | 1 | 2 | 1 |
| | 16053C>Y, 523-524delAC/non-del | 1 | 3 | 2 |
| | 16062A>R | 1 | 4 | 3 |
| | 16083C>Y, 172T>Y | 1 | 5 | 4 |
| | 16086T>Y | 1 | 6 | 5 |
| | 16126T>Y | 1 | 7 | 6 |
| | 16129G>R, 8C/9C/10C | 1 | 8 | 7 |
| | 16132A>R, 8C/9C | 1 | 9 | 8 |

| | | | | |
|---|---|---|---|---|
| | 16148C>Y | 1 | 10 | 9 |
| | 16151C>Y | 1 | 11 | 10 |
| | 16183A>R | 1 | 12 | 11 |
| | 16189T>Y | 1 | 13 | 12 |
| | 16192C>Y, 529G>R | 1 | 14 | 13 |
| | 16214C>Y | 1 | 15 | 14 |
| | 16254A>R | 1 | 16 | 15 |
| | 16266C>Y | 1 | 17 | 16 |
| | 16278C>Y | 1 | 18 | 17 |
| | 16327C>Y, 495C>Y | 1 | 19 | 18 |
| | 16345A>R, 7C/8C | 1 | 20 | 19 |
| | 16368T>C | 1 | 21 | 20 |
| | 16397T>Y | 2 | 22 | 21 |
| | 16457G>R, 124G>R | 1 | 23 | 22 |
| | 16546C>Y, 7C/8C | 1 | 24 | 23 |
| | 16567A>R | 1 | 25 | 24 |
| | 5A>R | 1 | 26 | 25 |
| | 26C>Y | 1 | 27 | 26 |
| | 29C>Y | 1 | 28 | 27 |
| | 31C>Y, 185G>R | 1 | 29 | 28 |
| | 57T>Y | 1 | 30 | 29 |
| | 71insG/non-ins | 1 | 31 | |
| | 103G>R | 1 | 32 | 30 |
| | 152T>Y | 1 | 33 | 31 |
| | 153A>R, 542C>Y, 7C/8C, 523-524delAC | 1 | 34 | 32 |
| | 204T>C | 1 | 35 | 33 |
| | 234A>R | 1 | 36 | 34 |
| | 354C>Y | 1 | 37 | 35 |
| | 367A>R | 1 | 38 | 36 |
| | 427C>Y, 8C/9C | 1 | 39 | 37 |
| | 437C>Y | 1 | 40 | 38 |
| | 471T>Y | 1 | 41 | 39 |
| | 503A>R | 1 | 42 | 40 |
| | 549C>Y | 1 | 43 | 41 |
| | 567A>R, 8C/9C | 1 | 44 | 42 |
| | 593T>Y | 1 | 45 | 43 |
| | 7C/8C | 5 | 46 | |
| | 8C/9C | 5 | 47 | |
| | 523-524delAC/non-del | 1 | 48 | |
| C-5 | aggregate sequence | 99 | 1 | |
| | 16086T>Y | 1 | 2 | 1 |
| | 16129G>R | 1 | 3 | 2 |
| | 16289A>R | 1 | 4 | 3 |
| | 16399A>R | 1 | 5 | 4 |
| | 16407C>Y | 1 | 6 | 5 |
| | 16497A>R | 1 | 7 | 6 |
| | 16519T | 1 | 8 | 7 |
| | 182C>Y | 1 | 9 | 8 |
| | 204T>C | 1 | 10 | 9 |
| | 204T>Y | 1 | 11 | 10 |
| | 251G>A | 1 | 12 | 11 |
| | 385A>G | 1 | 13 | 12 |
| | 513G>R | 1 | 14 | 13 |
| C-6 | aggregate sequence | 88 | 1 | |
| | 16380insC/non-ins | 1 | 2 | |
| | 36delG/non-del | 1 | 3 | |
| | 116A>R | 1 | 4 | 1 |

| | | | | |
|---|---|---|---|---|
| | 178A>R | 1 | 5 | 2 |
| | 189A>R | 1 | 6 | 3 |
| | 204T>C | 1 | 7 | 4 |
| | 524C>Y | 1 | 8 | 5 |
| C-7 | aggregate sequence | 53 | 1 | |
| | 16032T>Y, 460T>Y | 1 | 2 | 1 |
| | 16103A>R | 1 | 3 | 2 |
| | 16131T>Y, 16442C>Y | 1 | 4 | 3 |
| | 16149A>G | 1 | 5 | 4 |
| | 16277A>R | 1 | 6 | 5 |
| | 16282C>Y | 1 | 7 | 6 |
| | 16291C>Y, 8C/9C | 1 | 8 | 7 |
| | 16362T>Y | 1 | 9 | 8 |
| | 16363C>Y | 1 | 10 | 9 |
| | 16367A>R, 564G>R | 1 | 11 | 10 |
| | 16368T>T/G, 146T>Y | 1 | 12 | 11 |
| | 16462T>T/A, 227A>R | 1 | 13 | 12 |
| | 16502T>Y | 1 | 14 | 13 |
| | 16517G>R | 2 | 15 | 14 |
| | 16538C>Y, 8C/9C | 1 | 16 | 15 |
| | 71delG/non-del | 1 | 17 | |
| | 173T>Y | 1 | 18 | 16 |
| | 258C>Y | 1 | 19 | 17 |
| | 303C>Y | 1 | 20 | 18 |
| | 340C>Y | 1 | 21 | 19 |
| | 408T>Y | 1 | 22 | 20 |
| | 443A>R | 1 | 23 | 21 |
| | 497C>Y | 1 | 24 | 22 |
| | 517A>R | 1 | 25 | 23 |
| | 572C>Y | 1 | 26 | 24 |
| | 8C/9C, 573delC/non-del | 1 | 27 | |
| | 7C/8C | 4 | 28 | |
| | 8C/9C | 10 | 29 | |
| C-8 | aggregate sequence | 75 | 1 | |
| | 16122A>R, 228G>R | 1 | 2 | 1 |
| | 16129G>R | 1 | 3 | 2 |
| | 16135A>R | 1 | 4 | 3 |
| | 16139A>R | 1 | 5 | 4 |
| | 16159C>Y | 1 | 6 | 5 |
| | 16395C>Y | 1 | 7 | 6 |
| | 16399A>R | 1 | 8 | 7 |
| | 16399A>R, 16405A>R, 142T>Y | 1 | 9 | 8 |
| | 16399A>R, 16488C>Y | 1 | 10 | 9 |
| | 16417A>R, 16433A>R | 1 | 11 | 10 |
| | 16440T>Y | 1 | 12 | 11 |
| | 445C>Y | 1 | 13 | 12 |
| | 469C>Y | 1 | 14 | 13 |
| | 570C>Y | 1 | 15 | 14 |
| | 8C/9C | 3 | 16 | |
| ***Family D*** | | | | |
| D-1 | aggregate sequence | 57 | 1 | |
| | 16036delG/non-del, 180T>Y | 1 | 2 | 1 |
| | 16099C>Y, 15C>Y | 1 | 3 | 2 |
| | 16106G>R | 1 | 4 | 3 |
| | 16119A>R | 1 | 5 | 4 |
| | 16122A>R | 2 | 6 | 5 |

| | | | | |
|---|---|---|---|---|
| | 16129G>R | 1 | 7 | 6 |
| | 16130G>R | 1 | 8 | 7 |
| | 16132A>R, 16465C>Y | 1 | 9 | 8 |
| | 16189T>Y | 1 | 10 | 9 |
| | 16190C>Y, 7C/8C | 1 | 11 | 10 |
| | 16192C>Y, 16477G>R, 59T>Y, 7C/8C | 1 | 12 | 11 |
| | 16272A>R, 533A>R | 1 | 13 | 12 |
| | 16335A>R | 1 | 14 | 13 |
| | 16352T>Y, 438C>Y | 1 | 15 | 14 |
| | 16428G (same to CRS) | 2 | 16 | 15 |
| | 16439C>Y | 1 | 17 | 16 |
| | 16515A>R | 1 | 18 | 17 |
| | 109G>R, 526G>R, 533A>A/T | 1 | 19 | 18 |
| | 142T>Y | 1 | 20 | 19 |
| | 146T>Y, 456C>Y | 1 | 21 | 20 |
| | 234A>R, 6C/7C | 1 | 22 | 21 |
| | 242C>Y | 1 | 23 | 22 |
| | 244A>R | 1 | 24 | 23 |
| | 253C>Y, 356C>Y | 1 | 25 | 24 |
| | 262C>Y | 1 | 26 | 25 |
| | 260G>A | 1 | 27 | 26 |
| | 399T>Y, 506C>Y | 1 | 28 | 27 |
| | 490A>R | 1 | 29 | 28 |
| | 517A>R | 1 | 30 | 29 |
| | 540A>G | 1 | 31 | 30 |
| | 540A>R | 1 | 32 | 31 |
| | 561A>A/T | 1 | 33 | 32 |
| | 573delC/non-del | 2 | 34 | |
| | 523-524delAC/non-del | 1 | 35 | |
| | 6C/7C | 1 | 36 | |
| | 7C/8C | 1 | 37 | |
| | | | | |
| D-2 | aggregate sequence | 76 | 1 | |
| | 16051A>R, 16430A>A/T, 227A>R | 1 | 2 | 1 |
| | 16068T>Y, 40T>Y | 1 | 3 | 2 |
| | 16162A>R | 1 | 4 | 3 |
| | 16168C>Y, 16335A>R | 1 | 5 | 4 |
| | 16172T>Y | 1 | 6 | 5 |
| | 16192C>Y, 16435A>R | 1 | 7 | 6 |
| | 16397T>Y, 16547C>Y | 1 | 8 | 7 |
| | 16399A>R | 1 | 9 | 8 |
| | 16404C>Y | 1 | 10 | 9 |
| | 16408C>Y | 1 | 11 | 10 |
| | 47G>R | 1 | 12 | 11 |
| | 91C>Y | 1 | 13 | 12 |
| | 195T>Y | 1 | 14 | 13 |
| | 204T>C | 1 | 15 | 14 |
| | 204T>Y | 1 | 16 | 15 |
| | 419A>R | 1 | 17 | 16 |
| | 439A>R | 1 | 18 | 17 |
| | 490A>R | 1 | 19 | 18 |
| | 515A>A/T | 1 | 20 | 19 |
| | 523-524delAC/non-del | 1 | 21 | 20 |
| | | | | |
| ***Cord blood*** | | | | |
| CB-1 | aggregate sequence | 86 | 1 | |
| | 16134C>Y | 1 | 2 | 1 |
| | 16201C>Y | 1 | 3 | 2 |

| | | | | |
|---|---|---|---|---|
| | 194C>Y | 1 | 4 | 3 |
| | 247G>R | 1 | 5 | 4 |
| | 7C/8C | 3 | 6 | |
| | | | | |
| CB-2 | aggregate sequence (8C/9C) | 102 | 1 | |
| | 16022T>Y, 8C/9C | 1 | 2 | 1 |
| | 16038delA/non-del, 8C/9C/10C | 1 | 3 | |
| | 16100A>R, 8C/9C | 1 | 4 | 2 |
| | 16102T>Y, 265T>Y, 8C/9C | 1 | 5 | 3 |
| | 16111C>Y, 8C/9C | 1 | 6 | 4 |
| | 16179C>Y, 8C/9C | 1 | 7 | 5 |
| | 16215A>R, 16431C>Y, 8C/9C | 1 | 8 | 6 |
| | 16301C>Y, 8C/9C/10C | 1 | 9 | 7 |
| | 16432A>R, 8C/9C | 1 | 10 | 8 |
| | 16519T>Y, 8C/9C | 1 | 11 | 9 |
| | 85G>A, 200A>R, 8C/9C | 1 | 12 | 10 |
| | 86C>Y, 8C/9C | 1 | 13 | 11 |
| | 131T>Y, 8C/9C | 1 | 14 | 12 |
| | 143G>R, 8C/9C | 1 | 15 | 13 |
| | 152T>T/A, 8C/9C | 1 | 16 | 14 |
| | 166C>Y, 8C/9C/10C | 1 | 17 | 15 |
| | 178A>R, 8C/9C | 5 | 18 | 16 |
| | 178A>R, 8C/9C/10C | 2 | 19 | |
| | 193A>R, 8C/9C | 1 | 20 | 17 |
| | 308C>Y, 8C/9C/10C | 1 | 21 | 18 |
| | 416T>Y, 8C/9C | 1 | 22 | 19 |
| | 467C>Y, 8C/9C/10C | 1 | 23 | 20 |
| | 494C>Y, 8C/9C | 1 | 24 | 21 |
| | 498delC/non-del, 8C/9C | 1 | 25 | |
| | 559C>Y, 8C/9C | 1 | 26 | 22 |
| | 573delC/non-del, 8C/9C | 1 | 27 | |
| | 609C>Y, 8C/9C | 1 | 28 | 23 |
| | 8C/9C, 523-524delAC/non-del | 1 | 29 | |
| | 8C/9C/10C | 52 | 30 | |
| | | | | |
| CB-3 | aggregate sequence | 46 | 1 | |
| | 16274G>R | 1 | 2 | 1 |
| | 16278C>Y, 8C/9C | 1 | 3 | 2 |
| | 16343A>R, 16470G>R | 1 | 4 | 3 |
| | 16540C>Y, 8C/9C | 1 | 5 | 4 |
| | 16545T>Y, 8C/9C | 1 | 6 | 5 |
| | 5A>R | 1 | 7 | 6 |
| | 24A>R, 8C/9C | 1 | 8 | 7 |
| | 27C>Y | 2 | 9 | 8 |
| | 44C>Y, 8C/9C | 1 | 10 | 9 |
| | 72T>Y | 1 | 11 | 10 |
| | 116A>A/T, 229G>R | 1 | 12 | 11 |
| | 222C>Y | 1 | 13 | 12 |
| | 516C>Y | 1 | 14 | 13 |
| | 572C>Y | 1 | 15 | 14 |
| | 587C>Y | 1 | 16 | 15 |
| | 8C/9C | 29 | 17 | |
| | 8C/9C/10C | 4 | 18 | |

Note: For each donor, mutation in each single cell was scored relative to the aggregate sequence of all single cells, and mutation was numbered according to the revised Cambridge reference sequence. A site heterogeneous for both thymidine (T) and cytidine (C) was abbreviated as Y, and heterogeneous for adenosine (A) and guanosine (G) was abbreviated as R. A site heterogeneous for a transversion was highlighted by listing all status, e.g. 515A>A/T means site 515 has both A and T. We highlight the status of the length mutations of the C-tract at region 303-309 and the dinucleotide AC repeat at region 515-524 in these cells differ from the aggregate sequence by listing all the status, e.g. 8C means region 303-315 has CCCCCCCCTCCCCCC in the cell, 8C/9C means region 303-315 has a heteroplasmy of CCCCCCCCTCCCCCC and CCCCCCCCCTCCCCCC in the cell, 523-524delAC means this cell contains 4 repeats of AC at region 515-524, while 523-524insAC/non-ins means co-existence of 6 and 5 repeats of AC at region 515-524 in the cell. Back mutations relative to the sequence variation in aggregate sequence are denoted by adding "(same to CRS)" .