

Table S1 Mitochondrial genome sequences to test haplogroup assignment in MitoSuite

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| EU092714 | L0a1a1 |
| JX303911 | L0a1a2 |
| JQ044943 | L0a1a3 |
| EU092906 | L0a4 |
| HM771161 | L0a2a1 |
| EU092936 | L0b |
| EU092870 | L0f1 |
| KC345899 | L0k1a1 |
| KC345791 | L0d1a |
| KC345891 | L0d2d |
| DQ112737 | L1b1 |
| JQ705275 | L1c1a |
| AF346992 | L1c2a1a |
| HM771222 | L1c2b1a |
| EU273502 | L1c4a |
| EU273489 | L1c6 |
| JX303797 | L1c5 |
| EU092703 | L1c3a |
| EU273488 | L1c3b1a |
| EU273493 | L1c3c |
| DQ341060 | L5a1a |
| EF556173 | L5a1a |
| EU092888 | L5a1b |
| EU092943 | L5a1b |
| HM771198 | L5a1c |
| EU092699 | L5a2 |
| DQ341061 | L5b1a |
| EU092774 | L5b1b |
| DQ304928 | L2a1a |
| EU092761 | L2a1b |
| EU092747 | L2b1 |
| EU092661 | L2b3c |
| JQ044941 | L2c |
| AY195785 | L2c5 |
| JQ045050 | L2d |
| EU092794 | L2d1a |
| EU092724 | L2e |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| FJ460523 | L2e1a |
| EU092773 | L6a |
| EU092802 | L6a |
| DQ341063 | L6b |
| EU092686 | L6b |
| FJ460531 | L4a1 |
| DQ341064 | L4a1a |
| EU092935 | L4a2 |
| JQ044811 | L4b1a |
| EU092942 | L4b2a1 |
| EF184627 | L4b2a2 |
| EU092938 | L4b2a2a |
| JQ702504 | L4b2b1 |
| JN655813 | L3a1a |
| EU092726 | L3b1a |
| EU092891 | L3f1a |
| EU092660 | L3c |
| DQ112884 | L3d |
| EU092827 | L3e1 |
| DQ341069 | L3i1b |
| EU092822 | L3k |
| EF556171 | L3x1a1 |
| JN655830 | L3h1a1 |
| JQ702955 | M1a |
| AP012349 | M20 |
| KC505097 | M51a |
| DQ408676 | M3 |
| DQ408679 | M4 |
| AY922291 | M65a1 |
| EF556195 | M5a |
| HM156679 | M18a |
| JX289116 | M38 |
| HM346892 | M9a1 |
| AY519496 | C1a |
| FJ951462 | C4a1 |
| FJ951515 | C5a1 |
| FJ951594 | C7 |
| FJ383641 | C7b |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| FJ951440 | C5d1 |
| FJ951614 | C5c |
| FJ951472 | C5b1 |
| FJ951611 | C4e |
| GQ895155 | C4d |
| AY519493 | Z1 |
| FJ147318 | Z1a |
| AP008426 | Z2 |
| AP008841 | Z3 |
| EU597518 | Z3a1 |
| FJ383644 | Z3b |
| GU392051 | Z4 |
| AP013181 | Z4a1a1 |
| AP008553 | Z5 |
| FJ383629 | Z7 |
| GQ119027 | E1a |
| EF093539 | E1a1 |
| EF185804 | E1a1a1 |
| HQ700849 | E1a2 |
| HQ700847 | E1a2a |
| FJ428236 | E1b |
| EF061150 | E1b1 |
| HQ700856 | E2a |
| EF185816 | E2b1 |
| EF093542 | E2b2 |
| AP008845 | G1a1a4 |
| EF153773 | G1a1 |
| AY195762 | G1b |
| AY255137 | G1c |
| FJ198217 | G2a1 |
| FJ015040 | G2b1a |
| JF824842 | G2c |
| GU014566 | G3a1 |
| FJ748726 | G3b1 |
| AP008911 | G4 |
| DQ372885 | Q1 |
| AY289085 | Q1a |
| AF347003 | Q1b |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| EU597543 | Q1c |
| HQ113226 | Q2a |
| GQ214525 | Q2a1 |
| EF495218 | Q2b |
| AY289079 | Q3a |
| AY289089 | Q3a1 |
| EF061146 | Q3b |
| JQ704974 | D4 |
| JN253391 | D1 |
| AP011004 | D4b2 |
| AP008251 | D4c1b |
| AP008628 | D4d |
| AP008789 | D4e1a |
| EU660536 | D2a |
| JF824877 | D2b1 |
| AP008519 | D5c1a |
| AP011006 | D6a1 |
| JQ245777 | N1a1a |
| AY714031 | N5 |
| EU787451 | N2a |
| KC867130 | N3 |
| KC887495 | N7a1 |
| JX289118 | N8 |
| AP008726 | N9a1 |
| EF495214 | N13 |
| DQ112753 | N14 |
| JF739542 | N22 |
| AP010699 | A1 |
| DQ282395 | A2 |
| AY963575 | A6a |
| EF153771 | A12 |
| JF824996 | A15a |
| EF153780 | A16 |
| AP013217 | A3 |
| AP008265 | A5a |
| EU482363 | A8 |
| HM569228 | A10 |
| JQ245791 | I |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| HM454265 | I1a |
| FJ968796 | I1b |
| JQ705932 | I1c |
| EU570217 | I2 |
| JQ702041 | I3a |
| KJ021059 | I4 |
| JQ245724 | I5 |
| JQ705382 | I6a |
| KF146253 | I7 |
| AY289059 | O |
| DQ404447 | O1 |
| AY289056 | O1a |
| AF346963 | S1 |
| JN226144 | S1a |
| AY289051 | S2 |
| AY289066 | S3 |
| AY289062 | S4 |
| EF495220 | S5 |
| EU600318 | X1a |
| EF177437 | X3 |
| GQ200588 | X2 |
| DQ523631 | X2c |
| EU600325 | X2h |
| JQ245730 | X2i |
| FJ008043 | X2m1 |
| EF660942 | X2n |
| JQ245731 | X2o |
| HQ456226 | X4 |
| EU439939 | X2a1 |
| FJ825753 | X2a2 |
| EU935450 | X2j |
| FJ457949 | X2b |
| JQ705795 | X2d |
| KF056262 | W |
| JQ702793 | W1 |
| JQ705313 | W3a |
| KF146279 | W3b |
| KF146281 | W4 |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| KF146285 | W5 |
| JQ245723 | W6 |
| HM352797 | W7 |
| JQ245778 | W8 |
| JQ245759 | W9 |
| EF153813 | Y1 |
| HM776715 | Y1a |
| GU123044 | Y1b |
| AY255138 | Y1b1 |
| JF824992 | Y1b1a |
| AP008723 | Y2 |
| GQ119016 | Y2a |
| GQ119013 | Y2a1 |
| KC994134 | Y2a1a |
| AP008764 | Y2b |
| KC985149 | R1a |
| JQ705561 | R1a1a |
| KC985159 | R1a1 |
| HM030522 | R1a1b |
| KC985148 | R1a1c |
| KC985165 | R1b |
| JQ703633 | R1b1 |
| JX155266 | R2a |
| EF556167 | R2b |
| JX155271 | R2c |
| AY255136 | B6a |
| JX900371 | B4a1a1 |
| JQ704728 | B2a |
| AY519494 | B4b1a |
| AY255135 | B4d1 |
| KC733253 | B4e |
| JN857017 | B4j |
| KC521454 | B4c1a |
| EU597566 | B5a1a |
| JF824930 | B5b1a |
| HM357817 | F1a1a |
| AP010738 | F1c |
| AP012346 | F1f |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| KF849909 | F2 |
| AY255167 | F3a |
| AP013180 | F3b |
| AP008744 | F4a1a |
| AP013200 | F4a1b |
| JF824892 | F4a2 |
| AY289095 | F4b |
| JQ704041 | J2a1 |
| AY339579 | J2a1a1a1 |
| FJ348157 | J2a1a1a2 |
| JQ703568 | J2a1a1a3 |
| JQ705390 | J2a1a1b |
| JQ797924 | J2a2 |
| EF660967 | J2a2a |
| JQ797920 | J2a2a1 |
| JQ702563 | J2b1 |
| FJ213765 | J2b2 |
| AF347004 | P1 |
| DQ112897 | P2 |
| KC993994 | P10 |
| AY289052 | P3a |
| EF061158 | P4a |
| AY289063 | P5 |
| AY289053 | P6 |
| AY289054 | P7 |
| DQ404446 | P8 |
| KC993944 | P9 |
| KC405582 | T1a |
| JQ798058 | T2a1 |
| AY495273 | T2b |
| JQ798090 | T2c |
| AY714037 | T2d1a |
| EF177410 | T2e |
| JQ703997 | T2f |
| EU935442 | T2g1 |
| JN202724 | T2h |
| JX462725 | T3 |
| JQ702678 | R0a1 |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| HM185216 | R0a1a |
| HM185266 | R0a2 |
| HM185261 | R0a2b |
| HM185215 | R0a2c |
| JF717359 | R0a2d |
| HM185238 | R0a2h |
| HM185265 | R0a3 |
| JQ702940 | R0a4 |
| JF717361 | R0b |
| JQ705599 | HV0 |
| JF320654 | HV1 |
| AY713986 | HV2 |
| EF417833 | HV4 |
| EF419890 | HV5 |
| HQ658738 | HV6 |
| EU545443 | HV7 |
| EU545457 | HV8 |
| HM852849 | HV12a |
| JF700125 | HV13 |
| HQ384174 | H1 |
| EU597492 | H2a |
| AY738987 | H3 |
| EU935460 | H4 |
| AY495174 | H5 |
| HM765475 | H6a |
| AY495120 | H7 |
| AY738957 | H8a |
| EF660914 | H31 |
| HQ659700 | H100 |
| JQ702026 | V1a |
| JQ705658 | V2 |
| JQ703666 | V3 |
| AY495312 | V4 |
| AY339451 | V5 |
| FJ348207 | V6 |
| JQ705798 | V7 |
| JQ702025 | V8 |
| EF177445 | V9 |

| Accession | Haplogroup (PhyloTree) |
|------------------|-------------------------------|
| JQ704988 | V23 |
| HM852790 | U1a1 |
| KC521458 | U5 |
| EF064317 | U6a |
| JQ702004 | U2e2a4 |
| FN600416 | U2 |
| HM852891 | U3a |
| JQ703947 | U4a1 |
| AY882389 | U9a |
| AY714004 | U7 |
| JQ702759 | U8a |
| JQ706038 | K1a |
| DQ301795 | K1a1b1a |
| AY714044 | K1a1b2a |
| JQ705454 | K1a1c |
| JQ702168 | K1b1a1a |
| AY495241 | K2a |
| JX273249 | K2b |
| JQ704064 | K2b1b |
| DQ301796 | K2c |
| HM852886 | K3 |

Table S2 Detected heteroplasmic sites with MAF > 10 % in Avital et al., (2012).

| Sample ID | mtDNA Position | Region | Cell Type | Heteroplasmic fraction (Avital et al.) | Heteroplasmic fraction (MitoSuite) | > 10% (Avital et al.) | > 10% (MitoSuite) | Run |
|-----------|----------------|--------|-----------|--|------------------------------------|-----------------------|-------------------|-----------|
| 95451 | 9077 | ATP6 | Blood | 43.99 | 41.3 | ✓ | ✓ | SRR409202 |
| 95451 | 9077 | ATP7 | Muscle | 47.22 | 45.1 | ✓ | ✓ | SRR420854 |
| 95452 | 9077 | ATP7 | Blood | 38.53 | 48.9 | ✓ | ✓ | SRR420827 |
| 95452 | 9077 | ATP7 | Muscle | 41.82 | 28.7 | ✓ | ✓ | SRR420855 |
| 70251 | 16213 | D-LOOP | Blood | 10.64 | 15.2 | ✓ | ✓ | SRR420842 |
| 70251 | 16213 | D-LOOP | Muscle | 15.21 | 33.3 | ✓ | ✓ | SRR420957 |
| 70252 | 16213 | D-LOOP | Blood | 11.81 | 12.2 | ✓ | ✓ | SRR420844 |
| 68842 | 152 | D-LOOP | Muscle | 11.88 | 41.9 | ✓ | ✓ | SRR420859 |
| 68842 | 11299 | ND4 | Muscle | 11.52 | 11 | ✓ | ✓ | SRR420859 |
| 68842 | 14167 | ND6 | Muscle | 10.94 | 36 | ✓ | ✓ | SRR420859 |
| 68842 | 15132 | CYB | Muscle | 48.71 | 18.9 | ✓ | ✓ | SRR420859 |
| 68842 | 15132 | CYB | Blood | 48.11 | 39.5 | ✓ | ✓ | SRR420831 |
| 68841 | 14167 | CYB | Muscle | 12.42 | 47.2 | ✓ | ✓ | SRR420858 |
| 68841 | 15132 | D-LOOP | Blood | 41.96 | 21.3 | ✓ | ✓ | SRR420830 |
| 51402 | 385 | D-LOOP | Muscle | 10.3 | 7.5 | ✓ | - | SRR420863 |

Low-quality and duplicates reads were trimmed in QC procedures before we analyzed them using MitoSuite.