

Supplemental Data

A Bayesian Evaluation of Human Mitochondrial Substitution Rates

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Table S1. List of Published Date Estimates Used as Data Points in Figure 2

Study	Estimated Age of Human MRCA (kyr)	Calibration
Cann et al. (1987) ¹	143–285	Intraspecific
Vigilant et al. (1989) ²	476	Human-chimpanzee split
Hasegawa and Horai (1991) ³	280	Human-chimpanzee split
Vigilant et al. (1991) ⁴	166–249	Human-chimpanzee split
Nei (1992) ⁵	110–504	Human-chimpanzee split
Nei (1992) ⁵	760	Human-chimpanzee split
Pesole et al. (1992) ⁶	400	Human-chimpanzee split
Pesole et al. (1992) ⁶	600	Human-chimpanzee split
Stoneking et al. (1992) ⁷	133	Intraspecific
Stoneking et al. (1992) ⁷	137	Intraspecific
Hasegawa et al. (1993) ⁸	211	Human-chimpanzee split
Ruvolo et al. (1993) ⁹	195	Human-chimpanzee split
Ruvolo et al. (1993) ⁹	298	Human-chimpanzee split
Ruvolo et al. (1993) ⁹	506	Human-chimpanzee split
Horai et al. (1995) ¹⁰	450	Human-orangutan split
Horai et al. (1995) ¹⁰	143	Human-orangutan split
Wills (1995) ¹¹	436	Human-chimpanzee split
Wills (1995) ¹¹	806	Human-chimpanzee split
Krings et al. (1997) ¹²	120–150	Human-chimpanzee split
Parsons et al. (1997) ¹³	6.5	Pedigree rate
Krings et al. (1999) ¹⁴	163	Human-chimpanzee split
Arnason et al. (2000) ¹⁵	170	Human-chimpanzee split
Arnason et al. (2000) ¹⁵	400	Human-chimpanzee split
Ingman et al. (2000) ¹⁶	171.5	Human-chimpanzee split
Ingman and Gyllensten (2001) ¹⁷	167–171	Human-chimpanzee split
Tang et al. (2002) ¹⁸	214	Human-chimpanzee split
Mishmar et al. (2003) ¹⁹	198	Human-chimpanzee split
Kivisild et al. (2006) ²⁰	160	Human-chimpanzee split

Table S2. List of GenBank Accession Numbers for Sequences Used in This Study

Haplogroup	GenBank Accession	Haplogroup	GenBank Accession
Chimpanzee	X93335	M39	AY922275
Chimpanzee	CHPMTB	M40	AY922294
E	Pending, Soares et al. (in press) ²¹	M5	AY922292
E	Pending, Soares et al. (in press) ²¹	M5a	AY922260
H1	AY738972	M5a	AY922285
H1	AY738981	M5a	AY922289
H1	AY738982	M6	AY922296
H1	AY738973	M6	AY922307
H1	AY738974	Mstar	AY922302
H1	AY738975	Mstar	AY922299

H1	AY738976	Mstar	AY922300
H1	AY738977	Mstar	AY922253
H1	AY738978	Mstar	DQ404442
H1	AY738979	Mstar	DQ404443
H1	AY738980	N12a	AY289058
H3	AY738984	N12b	AY289059
H3	AY738985	N13	EF495214
H3	AY738986	N1d	AY714008
H3	AY738987	N5	AY714031
H3	AY738988	O	DQ404447
H3	AY738989	P1a1a1b	AY289087
H3	AY738990	P1a1a2	AY289092
H3	AY738991	P1a2	AY289086
H3	AY738992	P1b1	AY289084
I1	AY714041	P2	EF495215
L0	AF346985	P2a	AY289076
L0	AF346998	P2a	AY289080
L0	AF346999	P2a2	AY289052
L0	AF347008	P2b	AY289091
L0	AF347009	P3	AY289083
L1	AF346968	P3a	AY289053
L1	AF346969	P3b	AY289055
L1	AF346986	P4a	AY289057
L1	AF346987	P4b2	AY289064
L1	AF346992	P5	AY289063
L1	AF346996	P6	AY289054
L1	AF346997	P8	EF495221
L1	AY195780	P9	DQ404446
L1a2	AY195777	Q	EF495218
L1b1	AY195789	Q	AY289085
L1b2	AY195783	Q1	AY289077
L2	AF346976	Q1a1	AY289090
L2	AF346977	Q1a2a	AY289082
L2	AF346995	Q1a2b	AY289075
L2a	AY195776	Q2a	AY956412
L2a1	AY195788	Q2b1	AY289079
L2b	AY195766	Q2b2	AY289089
L2c	AY195785	Q2c	AY956414
M13	EF495219	Q3	AY289081
M14	EF495222	R1	AY714045
M2	AY922305	R14	EF495216
M27	DQ137411	R2	AY714007
M27a	DQ137410	R30	AY714001
M27b	DQ137402	R30	AY714032
M27b	DQ137403	R30	AY714047
M27b	DQ137404	R30	AY714050
M27c	DQ137405	R31	AY714046
M27c	DQ137406	R5	AY713983
M28a	DQ137400	R5	AY713984
M28a	DQ137401	R5	AY713985
M28b	DQ137398	R5	AY713991
M28b	DQ137399	R5	AY713993
M29	DQ137408	R6	AY713994
M29	DQ137409	R6	AY714028
M29	DQ137407	R7	AY714024
M29	EF495217	R7	AY714030
M2a	AY922306	R8	AY714009
M3	AY922263	R8	AY714011
M3	AY922266	S1	DQ404441
M31a1a	AY950298	S1	DQ404440

M31a1b	Pending, Barik et al. (in press) ²²	S1a	AF346963
M31a1b1	Pending, Barik et al. (in press) ²²	S2	AY289051
M31a1b2	AY950293	S2	AY289060
M31a1b2a	AY950294	S2d	AY289061
M31a2	EF060263	S4	EF495220
M31a2a1	EF060264	S4	AY289062
M31a2a2	EF060262	S4	AY289066
M31a2a3	EF060265	U1a	AY714038
M31b	N/A, Palanichamy et al. (2004) ²³	U2a	AY713990
M32a	AY950296	U2a	AY713992
M32b	DQ149518	U2b	AY714020
M32b	AY950295	U2b	AY714025
M33	AY922276	U2c	AY714010
M34	AY922304	U2e	AY714026
M34	AY922274	U3	AY714023
M35	AY922279	U7	AY714004
M35	AY922272	W1	AY714018
M36	AY922284	W1c	AY714039
M39	AY922269	W3	AY714043
M39	AY922288		

Table S3. List of Codons Classified as Being under Selection at a Significance Level of 5%, with Corresponding p Values

Gene	Codon Number	CRS Sites	dN:dS	p Value
<i>Positively Selected Codons</i>				
ND2	2306	5454–5456	5.00	0.017
<i>Negatively Selected Codons</i>				
ATP8	239	8402–8404	-3.12	0.033
COX1	366	6162–6164	-3.12	0.033
<i>COX1</i>	385	6219–6221	-3.00	0.037
<i>COX1</i>	398	6258–6260	-3.13	0.033
<i>COX1</i>	551	6717–6719	-3.00	0.037
<i>COX1</i>	556	6732–6734	-3.13	0.033
<i>COX1</i>	587	6825–6827	-3.00	0.037
COX1	757	7335–7337	-3.00	0.038
COX2	981	8147–8149	-3.00	0.037
COX2	1014	8246–8248	-6.00	0.014
COX3	1202	9747–9749	-4.18	0.011
<i>CYTB</i>	1402	15107–15109	-3.00	0.037
<i>CYTB</i>	1625	15776–15778	-3.00	0.037
<i>ND2</i>	2121	4899–4901	-3.00	0.037
<i>ND2</i>	2201	5139–5141	-3.00	0.037
<i>ND2</i>	2231	5229–5231	-4.00	0.012
<i>ND4</i>	2822	11903–11905	-4.00	0.013
<i>ND4L</i>	2969	10677–10679	-3.00	0.037
<i>ND5</i>	3018	12403–12405	-4.00	0.012
<i>ND5</i>	3173	12868–12870	-4.00	0.012
<i>ND5</i>	3220	13009–13011	-4.00	0.012
<i>ND5</i>	3307	13270–13272	-3.00	0.037
<i>ND5</i>	3380	13489–13491	-4.00	0.012

Supplemental References

1. Cann RL, Stoneking M, Wilson AC (1987) Mitochondrial DNA and human evolution. *Nature* 325:31–36.
2. Vigilant L, Pennington R, Harpending H, Kocher TD, Wilson AC (1989) Mitochondrial DNA sequences in single hairs from a southern African population. *Proc Natl Acad Sci USA* 86:9350–9354.
3. Hasegawa M, Horai S (1991) Time of the deepest root for polymorphism in human mitochondrial DNA. *J Mol Evol* 32:37–42.
4. Vigilant L, Stoneking M, Harpending H, Hawkes K, Wilson AC (1991) African populations and the evolution of human mitochondrial DNA. *Science* 253:1503–1507.
5. Nei M (1992) Age of the common ancestor of human mitochondrial DNA. *Mol Biol Evol* 9:1176–1178.
6. Pesole G, Sbisà E, Preparata G, Saccone C (1992) The evolution of the mitochondrial D-loop region and the origin of modern man. *Mol Biol Evol* 9:587–598.
7. Stoneking M, Sherry ST, Redd AJ, Vigilant L (1992) New approaches to dating suggest a recent age for human mtDNA ancestor. *Phil Trans R Soc Lond B* 337:167–175.
8. Hasegawa M, Di Rienzo A, Kocher TD, Wilson AC (1993) Toward a more accurate time scale for the human mitochondrial DNA tree. *J Mol Evol* 37:347–354.
9. Ruvolo M, Zehr S, von Dornum M, Pan D, Chang B, Lin J (1993) Mitochondrial COII sequences and modern human origins. *Mol Biol Evol* 10:1115–1135.
10. Horai S, Hayasaka K, Tsugane K, Takhata N (1995) Recent African origin of modern humans revealed by complete sequences of hominoid mitochondrial DNAs. *Proc Natl Acad Sci USA* 92:532–536.
11. Wills C (1995) When did Eve live? An evolutionary detective story. *Evolution* 49:593–607.
12. Krings M, Stone AC, Schmitz RW, Krainitzki H, Stoneking M, Pääbo S (1997) Neandertal DNA sequences and the origin of modern humans. *Cell* 90:19–30.
13. Parsons TJ, Muniec DS, Sullivan K, Woodyatt N, Alliston-Greiner R, Wilson MR, Berry DL, Holland KA, Weedn VW, Gill P, et al (1997) A high observed substitution rate in the human mitochondrial DNA control region. *Nat Genet* 15:363–368.
14. Krings M, Geisert H, Schmitz RW, Krainitzki H, Pääbo S (1999) DNA sequence of the mitochondrial hypervariable region II from the neandertal type specimen. *Proc Natl Acad Sci USA* 96:5581–5585.
15. Arnason U, Gullberg A, Burguete AS, Janke A (2000) Molecular estimates of primate divergences and new hypotheses for primate dispersals and the origin of modern humans. *Hereditas* 133:217–28.
16. Ingman M, Kaessmann H, Paabo S, Gyllensten U (2000) Mitochondrial genome variation and the origin of modern humans. *Nature* 408:708–713.
17. Ingman M, Gyllensten U (2001) Analysis of the complete human mtDNA genome: methodology and inferences for human evolution. *J Hered* 92:454–461.
18. Tang H, Siegmund DO, Shen P, Oefner PJ, Feldman MW (2002) Frequentist estimation of coalescence times from nucleotide sequence data using a tree-based partition. *Genetics* 161:447–459.
19. Mishmar D, Ruiz-Pesini E, Golik P, Macaulay V, Clark AG, Hosseini S, Brandon M, Easley K, Chen E, Brown MD, et al (2003) Natural selection shaped regional mtDNA variation in humans. *Proc Natl Acad Sci USA* 100:171–176.
20. Kivisild T, Shen P, Wall DP, Do B, Sung R, Davis K, Passarino G, Underhill PA, Scharfe C, Torroni A, et al (2006) The role of selection in the evolution of human mitochondrial genomes. *Genetics* 172:373–387.
21. Soares P, Trejaut JA, Loo J-H, Hill C, Mormina M, Lee C-L, Chen Y-M, Hudjashov G, Forster P, Macaulay V, Bulbeck D, Oppenheimer S, Lin M, and Richards MB (2008) Climate change and post-glacial human dispersals in Southeast Asia. *Molecular Biology and Evolution*, in press.
22. Barik SS, Sahani R, Prasad BVR, Endicott P, Metspalu M, Bhattacharya S, Sarkar BN, Annapoorna PCH, Sreenath J, Sun D, Sanchez JJ, Ho SYW, Chandrasekar A, and Rao VR. Detailed mtDNA genotypes permit a reassessment of the settlement and population structure of the Andaman Islands. *American Journal of Physical Anthropology*, in press.
23. Palanichamy MG, Sun C, Agrawal S, Bandelt HJ, Kong QP, Khan F, Wang CY, Chaudhuri TK, Palla V, Zhang YP. (2004). Phylogeny of mitochondrial DNA macrohaplogroup N in India, based on complete sequencing: implications for the peopling of South Asia. *American Journal of Human Genetics* 75:966-978.