AMERICAN JOURNAL OF PHYSICAL ANTHROPOLOGY 132:584–589 (2007)

Thomas Jefferson's Y Chromosome Belongs to a Rare European Lineage

Turi E. King, Georgina R. Bowden, Patricia L. Balaresque, Susan M. Adams, Morag E. Shanks, and Mark A. Jobling*

Department of Genetics, University of Leicester, University Road, Leicester LE1 7RH, UK

KEY WORDS Y chromosome; Thomas Jefferson; haplogroup K2

ABSTRACT We have characterized the Y chromosome carried by President Thomas Jefferson, the general rarity of which supported the idea that he, or a patrilineal relative, fathered the last son of his slave Sally Hemings. It belongs to haplogroup K2, a lineage representing only ~1% of chromosomes worldwide, and most common in East Africa and the Middle East. Phylogenetic network analysis of its Y-STR (short tandem repeat) haplotype shows that it is most closely related to an Egyptian K2 haplotype, but the presence of scattered and diverse European haplotypes within the network is nonetheless consistent with Jefferson's patrilineage belonging to an ancient and rare indigenous European type. This is supported by the observation that two of 85 unrelated British men sharing the surname Jefferson also share the President's Y-STR haplotype within haplogroup K2. Our findings represent a cautionary tale in showing the difficulty of assigning individual ancestry based on a Y-chromosome haplotype, particularly for rare lineages where population data are scarce. Am J Phys Anthropol 132:584–589, 2007.

Genetic evidence (Foster et al., 1998) has been used to support the idea that President Thomas Jefferson fathered Eston Hemings Jefferson (b. 1808), the last child of his slave Sally Hemings. The analysis exploited the paternally inherited Y chromosome and showed that a living patrilineal descendant of Eston shared a Y-chromosome haplotype with five patrilineal descendants of Thomas's paternal uncle. This suggested that either Thomas Jefferson himself, or one of his patrilineal relatives (Foster et al., 1999), was Eston's father. An important factor increasing the strength of this evidence was the general rarity of the 'Jefferson Y chromosome': in particular, the haplotype defined by Y-chromosomal STRs (short tandem repeats) was not observed in >1200 chromosomes worldwide (Foster et al., 1998). In the eight years since this study, Y-chromosomal haplotyping has improved greatly in resolution, and the amount of available comparative data has grown enormously. We now revisit the Jefferson Y chromosome, and show that, despite these developments, it remains of an extremely unusual type. Although membership of a rare and diverse Y-chromosomal lineage makes assignment to a particular population of origin difficult, we show that Jefferson's Y chromosome has a likely origin in western Europe.

MATERIALS AND METHODS

Samples and comparative data

The Virginia Jefferson samples, originally described in (Foster et al., 1998), were obtained and kindly made available by Gene Foster. French samples were a gift from André Chaventré, Christelle Richard, and Jean-Paul Moisan. British samples were from the collections of the authors.

Haplogroup data on 14,685 chromosomes were taken from the literature (Semino et al., 2000; Cruciani et al., 2002; Semino et al., 2002; Al-Zahery et al., 2003; Capelli et al., 2003; Arredi et al., 2004; Cinnioglu et al., 2004; Flores et al., 2004; Luis et al., 2004; Shen et al., 2004; Gonçalves et al., 2005; Hurles et al., 2005; Karafet et al., 2005; Sanchez et al., 2005; Beleza et al., 2006; Hammer et al., 2006a; Hammer et al., 2006b; Sengupta et al., 2006).

Y-chromosome haplotyping

Haplogroup (hg) K2 chromosomes are defined by possession of the derived state of the M70 A-to-C single-nucleotide polymorphism (SNP). They were identified in the Virginia Jefferson DNAs, and in British, French, and Iberian Peninsula samples, by typing M70 using PCR-RFLP (polymerase chain reaction restriction fragment length polymorphism) analysis. A 257-bp product containing the M70 SNP was amplified using published M70 oligonucleotide primers (Underhill et al., 2001; Y Chromosome Consortium, 2002) and digested with the restriction enzyme *Psr*I (Sibenzyme), which cleaves the ancestral (A) allele only, prior to resolution by agarose gel electrophoresis.

British and French hgK2 chromosomes were analyzed with 17 Y-STRs, typed according to Bosch et al. (2002), and combined with 8-locus Y-STR haplotypes (DYS19, DYS389I, DYS389II-I, DYS390, DYS391, DYS392, DYS393, DYS439) from Iberian Peninsula (Bosch et al., 2002), Turkish (Cinnioglu et al., 2004), Omani, Egyptian (Luis et al., 2004), Somali (Sanchez et al., 2005), and Portuguese (Beleza et al., 2006) data to construct a median joining network, displaying the phylogenetic relationships between

Grant sponsors: The Wellcome Trust, Medical Research Council (MRC).

*Correspondence to: Prof. Mark A. Jobling, Department of Genetics, University of Leicester, University Road, Leicester LE1 7RH, UK. E-mail: maj4@leicester.ac.uk

Received 3 July 2006; accepted 4 December 2006

DOI 10.1002/ajpa.20557

Published online 1 February 2007 in Wiley InterScience (www.interscience.wiley.com).

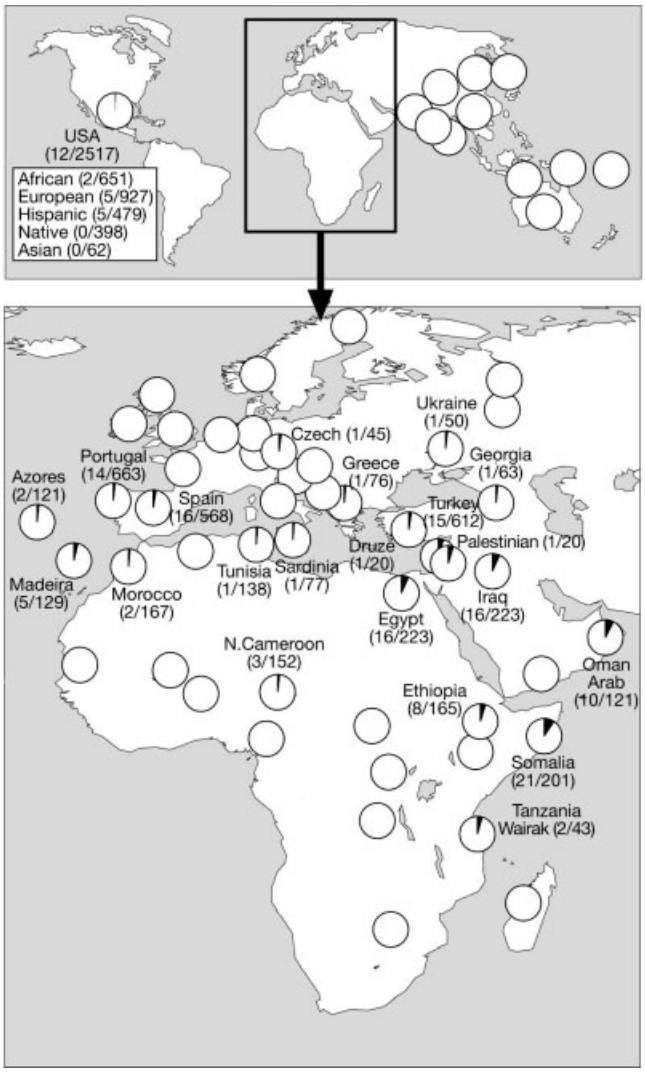

Fig. 1. Worldwide distribution of Y chromosomes belonging to haplogroup K2 and worldwide distribution of hgK2 among 14,685 chromosomes taken from the literature (see Materials and Methods section for sources) are shown. The black sector of each pie chart is proportional to the frequency of hgK2. Populations in which hgK2 is present are named, and the accompanying numbers (e.g. 16/223) indicate number of K2 chromosomes/total sample size. British and French cases identified in this study are not included. The upper panel includes the frequencies of hgK2 in subpopulations of the USA (Hammer et al., 2006b).

haplotypes (Bandelt et al., 1999), within the program Network 4.1.0.9 (www.fluxus-engineering.com/sharenet.htm/).

To resolve extensive reticulations from the network, weighting was performed according to Qamar et al. (2002). Variance in all K2 chromosomes was calculated for each Y-STR locus, loci with high variance being given low weights, and vice versa. Time-to-most-recent-common-ancestor (TMRCA) was estimated within Network from the rho statistic (the mean number of mutations from the assumed root of the network), using a 25-year generation time and a mean per-locus, per-generation mutation rate of 6.9×10^{-4} (Zhivotovsky et al., 2004).

DNA from unrelated British men named Jefferson (82 individuals), Jeffreson (two), or Jeaffreson (one) was haplotyped using 17 Y-STRs and 10 Y-chromosomal binary markers (King et al., 2006), plus the SNP M70 as described above. Full haplotype data are available from the authors. Network construction was as above, with weighting based on Y-STR variance observed in 291 British hgR1b chromosomes. Binary markers were included in network construction (at a weight of 99) to preserve the phylogenetic relationship of haplogroups within the network.

The minisatellite MSY1, which contains a hypervariable array of 25-bp repeat units, was typed by MVR-PCR (Jobling et al., 1998) to map the position of variant repeats within the array.

TMRCA between the British hgK2 Jefferson haplotype and the commonest 17-locus Virginia Jefferson haplotype was estimated using a Bayesian approach as described (Walsh, 2001; King et al., 2006).

RESULTS

Thomas Jefferson's own family tradition traced his ancestry to Wales (Brodie, 1974), so we might expect the Y chromosome carried by the six Jefferson descendants to belong to a typical western European haplogroup (hg). Previous analysis (Foster et al., 1998) had placed it with poor resolution in part of the Y chromosome phylogeny defined in current nomenclature as hgBK(xP). To refine this, we typed additional binary markers, obtaining the surprising result that the Jefferson chromosome belongs to hgK2, defined by the derived state of the SNP M70. This hg is rare worldwide: in 14,685 chromosomes in the literature that have been surveyed (see Materials and Methods section), only 150 examples (1.02%) are reported. Figure 1 shows their global distribution; apart from 12 cases from non-native populations of the USA, they are confined to Africa and west Eurasia. They reach their highest frequencies in the Middle East and northern East Africa, exceeding 7% in Somalia, Oman, Egypt, and Iraq, but are also found at lower frequency in the central and western Mediterranean, including the Iberian Peninsula. In a sample of 1,772 chromosomes from the British Isles (Capelli et al., 2003), only a single chromosome, defined as hgK(xP,N3), is a candidate for hgK2. We typed the M70 polymorphism in a sample of 421 British males and found two hgK2 chromosomes (0.5%).

To address the geographical origins of the Jefferson Y, we first compared its 9-locus Y-STR haplotype with a global online database of 13,083 9-locus haplotypes (Roewer et al., 2001); it finds no matches. We next compared it with haplotypes of other K2 chromosomes from the literature, adding further examples that we identified from the Iberian Peninsula (13/987; 1.3%), western France (3/561; 0.5%), and Britain (Table 1), typed as described (Bosch et al., 2002). A phylogenetic network can be used to represent the relationships between the Y-STR haplotypes, and shows that the chromosomes are diverse (Fig. 2a), with an estimated TMRCA (time-to-most-recent-common-ancestor) of 20,700 $\pm$ 4100 years. The Jefferson haplotype lies at the periphery of the network; its nearest (one mutational step) neighbor is an Egyptian, while Iberian haplotypes lie only two steps away. Does this suggest a recent non-European origin? There is little geographical clustering in the network, apart from the Somali haplotypes, and the Europeans are scattered widely, with the three British haplotypes well separated from each other. This seems compatible with the British examples, including the Jefferson haplotype, being members of an ancient and diverse indigenous European lineage, rather than recent immigrants from the Middle East or Africa, although this is far from conclusive.

TABLE 1. *Y-STR haplotypes of haplogroup K2 chromosomes*

Name	*Population*	*DYS19*	*DYS388*	*DYS389I*	*DYS389II - I*	*DYS390*	*DYS391*	*DYS392*	*DYS393*	*DYS434*	*DYS435*	*DYS436*	*DYS437*	*DYS438*	*DYS439*	*DYS460*	*DYS461*	*DYS462*
J47[a]	Virginia Jefferson	15	12	12	15	24	10	15	13	11	11	12	14	9	11	10	11	13
J50	Virginia Jefferson	15	12	12	15	24	10	16	13	11	11	12	14	9	12	10	11	13
J49	Virginia Jefferson	15	12	12	15	24	10	15	13	11	11	12	14	9	12	10	11	13
J41	Virginia Jefferson	15	12	12	15	24	10	15	13	11	11	12	14	9	12	10	11	13
J42	Virginia Jefferson	15	12	12	15	24	10	15	13	11	11	12	14	9	12	10	11	13
H1	Virginia Jefferson	15	12	12	15	24	10	15	13	11	11	12	14	9	12	10	11	13
IP154	Iberian Peninsula	13	12	14	14	22	10	13	13	11	11	12	15	9	11	10	11	12,13
IP163	Iberian Peninsula	13	12	13	15	23	10	13	14	11	11	12	15	9	11	11	11	13
IP211	Iberian Peninsula	13	12	14	15	24	10	13	13	11	11	12	14	9	12	10	12	12
IP220	Iberian Peninsula	13	12	14	13	24	10	13	13	11	11	12	14	9	12	10	12	12
IP371	Iberian Peninsula	13	12	13	16	23	10	13	13	11	11	12	15	9	13	10	11	12
IP469	Iberian Peninsula	14	12	13	17	23	10	13	13	11	11	12	14	9	11	10	11	13
IP146	Iberian Peninsula	14	12	13	18	23	10	14	13	11	11	12	14	9	11	10	11	11
IP248	Iberian Peninsula	15	11	14	18	23	10	13	13	11	11	12	14	9	11	11	11	12
IP263	Iberian Peninsula	15	12	13	17	23	11	13	13	11	11	12	14	9	11	11	11	14
IP755	Iberian Peninsula	15	12	13	17	25	10	14	13	11	11	12	14	9	11	10	11	12
IP1023	Iberian Peninsula	15	12	12	17	24	10	14	13	11	11	12	14	9	11	10	11	13
IP427	Iberian Peninsula	15	12	13	16	23	10	15	13	11	11	12	14	9	10	10	11	13
IP572	Iberian Peninsula	16	12	12	16	23	10	14	13	11	11	12	14	9	9	10	11	12
W37	French	13	12	14	16	22	10	13	13	nd	nd	nd	15	9	11	10	nd	nd
W40	French	14	12	13	16	23	10	14	13	nd	nd	nd	14	9	11	11	nd	nd
V85	French	13	12	13	16	23	10	14	14	nd	nd	nd	15	9	11	11	nd	nd
GB647	British non-Jefferson	13	12	14	16	22	10	13	13	11	11	12	14	9	11	10	11	12
GB1229	British non-Jefferson	15	12	14	17	23	10	14	13	11	11	12	14	9	11	10	11	12

[a] Names as in Foster et al., 1988. nd: not determined.

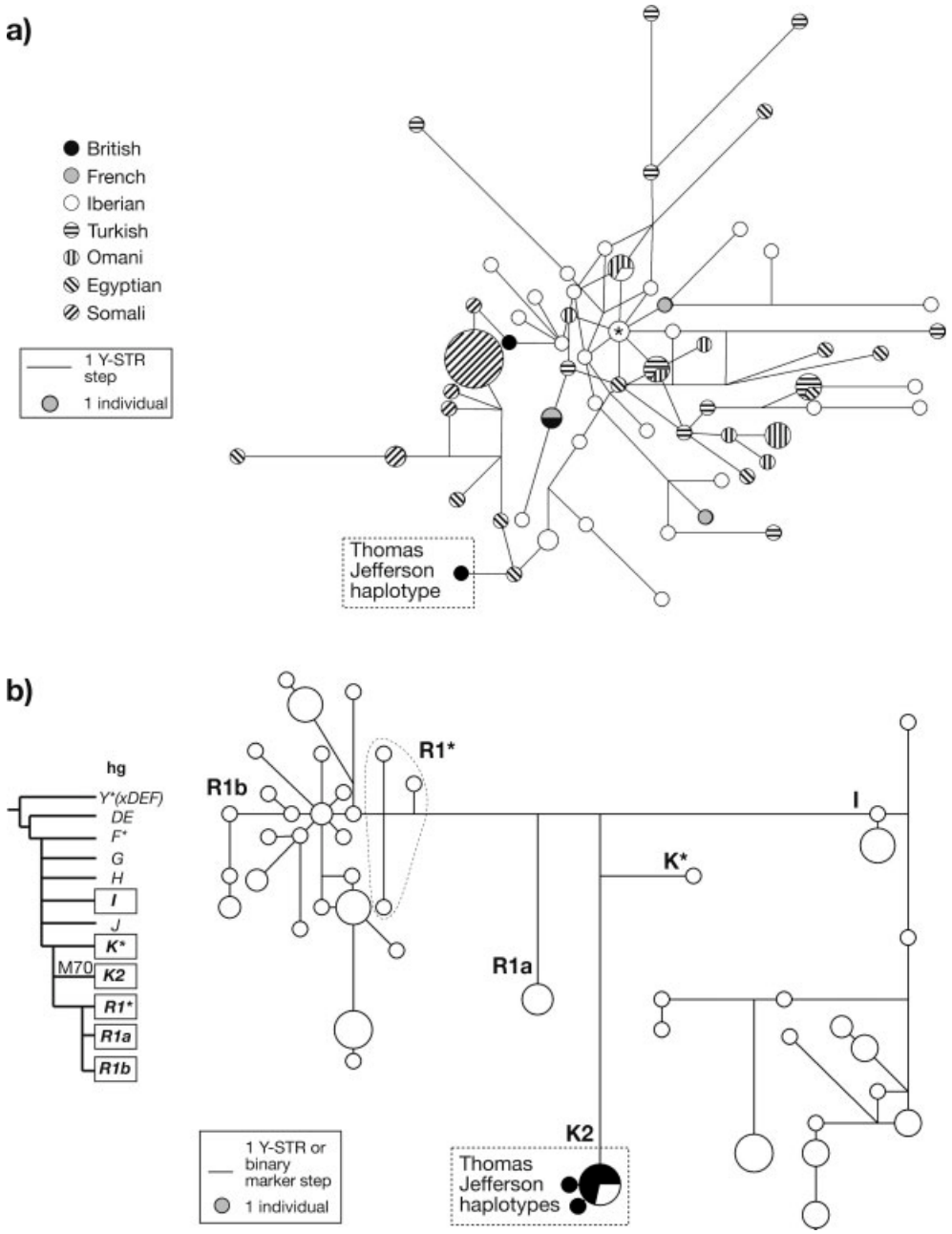

Fig. 2. Diversity of Y-STR haplotypes belonging to haplogroup K2, and within the Jefferson surname. (a) Weighted median joining network (Bandelt et al., 1999) containing the 8-locus Y-STR (DYS19, DYS389I, DYS389II-I, DYS390, DYS391, DYS392, DYS393, DYS439) haplotypes of 86 hgK2 chromosomes. Circles represent haplotypes, with area proportional to frequency and shaded or hatched according to population. The ancestral node used for dating is indicated by an asterisk. Data were taken from the literature: Iberian Peninsula (Bosch et al., 2002), Turkish (Cinnioglu et al., 2004), Omani, Egyptian (Luis et al., 2004), Somali (Sanchez et al., 2005), and Portuguese (Beleza et al., 2006), with the addition of British and French chromosomes identified and analyzed in this study. (b) Y-chromosomal haplotypes of men named Jefferson. Left: binary marker phylogeny of the Y chromosome, showing haplogroup (hg) names (Y Chromosome Consortium, 2002). Right: weighted median joining network containing the 17-locus Y-STR haplotypes of 91 Jefferson men including the six hgK2 Virginia Jeffersons (Foster et al., 1998). Circles represent haplotypes, with area proportional to frequency, and haplogroups labeled. The two hgK2 British Jefferson haplotypes are indicated by a white sector. Network construction was as in (a), with inclusion of binary markers to preserve phylogenetic information.

If we did not have prior knowledge about the ancestry of the Jefferson haplotype, we might assign it to an Egyptian origin.

Despite its general rarity, there remains a sizeable subpopulation in which Thomas Jefferson's Y chromosome could be frequent—all men named Jefferson. This is because both Y chromosomes and surnames are patrilinearly inherited, and a correlation exists between Y haplotypes and surnames (Sykes and Irven, 2000; King et al., 2006; McEvoy and Bradley, 2006). To test this, we randomly recruited 85 unrelated British Jefferson males from the total population of ~2,100 men carrying this surname, and typed their Y

chromosomes using 11 binary markers and 17 Y-STRs (King et al., 2006). Diverse haplotypes are distributed among six haplogroups (Fig. 2b), consistent with multiple founders for the name, and/or historical nonpaternities. However, two of the men (~2%; males GB1078 and GB1151) belong to hgK2, and both carry the Thomas Jefferson Y-STR haplotype. Their paternal grandfathers were born in Yorkshire and the west Midlands respectively, and neither reports any known familial links to the USA. These men share recent paternal ancestry with the six Virginia Jefferson descendants described by Foster et al. (Foster et al., 1998). How long ago they shared an ancestor is difficult to estimate accurately, but given the identical Y-STR haplotypes, median TMRCA between the British hgK2 Jefferson haplotype and the commonest 17-locus Virginia haplotype can be calculated as 11 generations (with a very broad 95% confidence interval of 0.4–60 generations). A close relationship is supported by the structure of the fast-evolving Y-specific minisatellite MSY1 (Jobling et al., 1998) in these males. The Virginia Jefferson descendants (Foster et al., 1998) share the MSY1 structure $(3)_5(1)_{14}(3)_{32}(4)_{16}$ (the number in parenthesis referring to the repeat type, and the subscript the number of repeats in each successive block); one of the two British Jeffersons has exactly the same MSY1 structure, while the other has a single repeat-unit deletion $[(3)_5(1)_{14}(3)_{32}(4)_{15}]$. Presence of the Jefferson haplotype in these apparently unrelated British men supports the idea that the President's recent paternal ancestry is in western Europe, rather than the Middle East.

DISCUSSION

Globally, many Y chromosomes belong to lineages with well-defined distributions, and currently exist in large numbers of males, sometimes regarded as patrilineal 'clans' (Oppenheimer, 2003). One striking case is the highly homogeneous cluster within hgC*(xC3c) representing ~0.5% of Y chromosomes worldwide, and interpreted as a product of the dynasty of Genghis Khan (Zerjal et al., 2003); another is the common sublineage within hgR1b3 originating in Ireland, and ascribed to the *Uí Néill* dynasty (Moore et al., 2006), which is carried by 2–3 million men. Nevertheless, a minority of Y chromosome types, of which the Jefferson Y is an example, remains highly distinctive and scarce, belonging to diverse lineages with poorly defined population distributions.

This study represents a cautionary tale, since the interpretation of the origins of rare and unusual lineages such as that of Thomas Jefferson can be heavily biased by how they are defined, and by what comparative data are available. In this particular case, the ancestry of the individual in question was known, but without this knowledge, and using data available from the literature, it is likely that he would have been assigned Egyptian or Middle Eastern ancestry. Our finding of two other men from the general British population with hgK2 Y chromosomes, and in particular of two British Jefferson men who share the President's haplotype, reinforces the idea that Jefferson's recent paternal ancestry is European, though this conclusion could in principle change with more extensive sampling in the future.

In the modern era of 'genetic genealogy', many companies offer to provide information about individual ancestry. Despite an increasing and sometimes impressive degree of genetic literacy among clients, the inherent lack of power of individual ancestry prediction based on a single genetic locus is often not appreciated, and understandably is not emphasized by providers of testing services.

The popularity of genetic testing has caused the public fascination with Thomas Jefferson to extend beyond the man to the details of his Y chromosome haplotype: one DNA-typing company offers a 'Famous Y-DNA' testing service, including matching to Jefferson, while the internet is rich with speculations about his ancestry. Our findings show that firm conclusions can be difficult to draw, but taken together are consistent with Jefferson's patrilineage belonging to an ancient and rare indigenous west European type; they also increase the strength of the evidence that Thomas Jefferson, or one of his patrilineal relatives, rather than some unrelated man, was the father of Eston Hemings Jefferson.

ACKNOWLEDGMENTS

We thank all DNA donors, Juan Sanchez, Sandra Beleza and Rene Herrera for sharing haplotype data, André Chaventré, Christelle Richard and Jean-Paul Moisan for access to French samples, and Gene Foster for obtaining and making available the Virginia Jefferson samples. We are grateful to two anonymous reviewers for helpful comments. T.E.K. was supported by a Wellcome Prize Studentship (061129), M.E.S. by the MRC, G.R.B., P.L.B., and S.M.A. by the Wellcome Trust and M.A.J. by a Wellcome Trust Senior Fellowship in Basic Biomedical Science (057559).

LITERATURE CITED

Al-Zahery N, Semino O, Benuzzi G, Magri C, Passarino G, Torroni A, Santachiara-Benerecetti AS. 2003. Y-chromosome and mtDNA polymorphisms in Iraq, a crossroad of the early human dispersal and of post-Neolithic migrations. Mol Phylogenet Evol 28:458–472.

Arredi B, Poloni ES, Paracchini S, Zerjal T, Fathallah DM, Makrelouf M, Pascali VL, Novelletto A, Tyler-Smith C. 2004. A predominantly neolithic origin for Y-chromosomal DNA variation in North Africa. Am J Hum Genet 75:338–345.

Bandelt H-J, Forster P, Röhl A. 1999. Median-joining networks for inferring intraspecific phylogenies. Mol Biol Evol 16:37–48.

Beleza S, Gusmao L, Lopes A, Alves C, Gomes I, Giouzeli M, Calafell F, Carracedo A, Amorim A. 2006. Micro-phylogeographic and demographic history of Portuguese male lineages. Ann Hum Genet 70:181–194.

Bosch E, Lee AC, Calafell F, Arroyo E, Henneman P, de Knijff P, Jobling MA. 2002. High resolution Y chromosome typing: 19 STRs amplified in three multiplex reactions. Forensic Sci Int 125:42–51.

Brodie FM. 1974. Thomas Jefferson: an intimate history. London: Eyre Methuen.

Capelli C, Redhead N, Abernethy JK, Gratrix F, Wilson JF, Moen T, Hervig T, Richards M, Stumpf MPH, Underhill PA, Bradshaw P, Shaha A, Thomas MG, Bradman N, Goldstein DB. 2003. A Y chromosome census of the British Isles. Curr Biol 13:979–984.

Cinnioglu C, King R, Kivisild T, Kalfoglu E, Atasoy S, Cavalleri GL, Lillie AS, Roseman CC, Lin AA, Prince K, Oefner PJ, Shen P, Semino O, Cavalli-Sforza LL, Underhill PA. 2004. Excavating Y-chromosome haplotype strata in Anatolia. Hum Genet 114: 127–148.

Cruciani F, Santolamazza P, Shen PD, Macaulay V, Moral P, Olckers A, Modiano D, Holmes S, Destro-Bisol G, Coia V, Wallace DC, Oefner PJ, Torroni A, Cavalli-Sforza LL, Scozzari R, Underhill PA. 2002. A back migration from Asia to sub-Saharan Africa is supported by high-resolution analysis of human Y-chromosome haplotypes. Am J Hum Genet 70:1197–1214.

Flores C, Maca-Meyer N, Gonzalez AM, Oefner PJ, Shen P, Perez JA, Rojas A, Larruga JM, Underhill PA. 2004. Reduced genetic structure of the Iberian peninsula revealed by Y-chromosome analysis: implications for population demography. Eur J Hum Genet 12:855–863.

Foster EA, Jobling MA, Taylor PG, Donnelly P, de Knijff P, Mieremet R, Zerjal T, Tyler-Smith C. 1998. Jefferson fathered slave's last child. Nature 396:27–28.

Foster EA, Jobling MA, Taylor PG, Donnelly P, de Knijff P, Mieremet R, Zerjal T, Tyler-Smith C. 1999. The Thomas Jefferson paternity case. Nature 397:32.

Gonçalves R, Freitas A, Branco M, Rosa A, Fernandes AT, Zhivotovsky LA, Underhill PA, Kivisild T, Brehm A. 2005. Y-chromosome lineages from Portugal, Madeira and Acores record elements of Sephardim and Berber ancestry. Ann Hum Genet 69:443–454.

Hammer MF, Karafet TM, Park H, Omoto K, Harihara S, Stoneking M Horai S. 2006a. Dual origins of the Japanese: common ground for hunter-gatherer and farmer Y chromosomes. J Hum Genet 51:47–58.

Hammer MF, Chamberlain VF, Kearney VF, Stover D, Zhang G, Karafet T, Walsh B, Redd AJ. 2006b. Population structure of Y chromosome SNP haplogroups in the United States and forensic implications for constructing Y chromosome STR databases. Forensic Sci Int 164:45–56.

Hurles ME, Sykes BC, Jobling MA, Forster P. 2005. The dual origin of the Malagasy in island southeast Asia and East Africa: evidence from maternal and paternal lineages. Am J Hum Genet 76:894–901.

Jobling MA, Bouzekri N, Taylor PG. 1998. Hypervariable digital DNA codes for human paternal lineages: MVR-PCR at the Y-specific minisatellite, MSY1 (*DYF155S1*). Hum Mol Genet 7:643–653.

Karafet TM, Lansing JS, Redd AJ, Reznikova S, Watkins JC, Surata SP, Arthawiguna WA, Mayer L, Bamshad M, Jorde LB, Hammer MF. 2005. Balinese Y-chromosome perspective on the peopling of Indonesia: genetic contributions from pre-neolithic hunter-gatherers, Austronesian farmers, and Indian traders. Hum Biol 77:93–114.

King TE, Ballereau SJ, Schürer K, Jobling MA. 2006. Genetic signatures of coancestry within surnames. Curr Biol 16:384–388.

Luis JR, Rowold DJ, Regueiro M, Caeiro B, Cinnioglu C, Roseman C, Underhill PA, Cavalli-Sforza LL, Herrera RJ. 2004. The levant versus the horn of Africa: evidence for bidirectional corridors of human migrations. Am J Hum Genet 74: 532–544.

McEvoy B, Bradley DG. 2006. Y-chromosomes and the extent of patrilineal ancestry in Irish surnames. Hum Genet 119:212–219.

Moore LT, McEvoy B, Cape E, Simms K, Bradley DG. 2006. A Y-chromosome signature of hegemony in Gaelic Ireland. Am J Hum Genet 78:334–338.

Oppenheimer S. 2003. Out of Eden: the peopling of the world. London: Constable.

Qamar R, Ayub Q, Mohyuddin A, Helgason A, Mazhar K, Mansoor A, Zerjal T, Tyler-Smith C, Mehdi SQ. 2002. Y-chromosomal DNA variation in Pakistan. Am J Hum Genet 70:1107–1124.

Roewer L, Krawczak M, Willuweit S, Nagy M, Alves C, Amorim A, Anslinger K, Augustin C, Betz A, Bosch E, Caglià A, Carracedo A, Corach D, Dobosz T, Dupuy BM, Füredi S, Gehrig C, Gusm~ao L, Henke J, Henke L, Hidding M, Hohoff C, Hoste B, Jobling MA, Kärgel HJ, de Knijff P, Lessig R, Liebeherr E, Lorente M, Martínez-Jarreta B, Nievas P, Nowak M, Parson W, Pascali VL, Penacino G, Ploski R, Rolf B, Sala A, Schmidt U, Schmitt C, Schneider PM, Szibor R, Teifel-Greding J, Kayser M. 2001. Online reference database of Y-chromosomal short tandem repeat (STR) haplotypes. Forensic Sci Int 118:103–111.

Sanchez JJ, Hallenberg C, Borsting C, Hernandez A, Morling N. 2005. High frequencies of Y chromosome lineages characterized by E3b1, DYS19-11, DYS392-12 in Somali males. Eur J Hum Genet 13:856–866.

Semino O, Passarino G, Oefner PJ, Lin AA, Arbuzova S, Beckman LE, De Benedictis G, Francalacci P, Kouvatsi A, Limborska S, Marcikiae M, Mika A, Mika B, Primorac D, Santachiara-Benerecetti AS, Cavalli-Sforza LL, Underhill PA. 2000. The genetic legacy of Paleolithic Homo sapiens sapiens in extant Europeans: a Y chromosome perspective. Science 290: 1155–1159.

Semino O, Santachiara-Benerecetti AS, Falaschi F, Cavalli-Sforza LL, Underhill PA. 2002. Ethiopians and Khoisan share the deepest clades of the human Y-chromosome phylogeny. Am J Hum Genet 70:265–268.

Sengupta S, Zhivotovsky LA, King R, Mehdi SQ, Edmonds CA, Chow CE, Lin AA, Mitra M, Sil SK, Ramesh A, Usha Rani MV, Thakur CM, Cavalli-Sforza LL, Majumder PP, Underhill PA. 2006. Polarity and temporality of high-resolution Y-chromosome distributions in India identify both indigenous and exogenous expansions and reveal minor genetic influence of Central Asian pastoralists. Am J Hum Genet 78:202–221.

Shen P, Lavi T, Kivisild T, Chou V, Sengun D, Gefel D, Shpirer I, Woolf E, Hillel J, Feldman MW, Oefner PJ. 2004. Reconstruction of patrilineages and matrilineages of Samaritans and other Israeli populations from Y-chromosome and mitochondrial DNA sequence variation. Hum Mutat 24:248–260.

Sykes B, Irven C. 2000. Surnames and the Y chromosome. Am J Hum Genet 66:1417–1419.

Underhill PA, Passarino G, Lin AA, Shen P, Lahr MM, Foley RA, Oefner PJ, Cavalli-Sforza LL. 2001. The phylogeography of Y chromosome binary haplotypes and the origins of modern human populations. Ann Hum Genet 65:43–62.

Walsh B. 2001. Estimating the time to the most recent common ancestor for the Y chromosome or mitochondrial DNA for a pair of individuals. Genetics 158:897–912.

Y Chromosome Consortium. 2002. A nomenclature system for the tree of human Y-chromosomal binary haplogroups. Genome Res 12:339–348.

Zerjal T, Xue Y, Bertorelle G, Wells RS, Bao W, Zhu S, Qamar R, Ayub Q, Mohyuddin A, Fu S, Li P, Yuldasheva N, Ruzibakiev R, Xu J, Shu Q, Du R, Yang H, Hurles ME, Robinson E, Gerelsaikhan T, Dashnyam B, Mehdi SQ, Tyler-Smith C. 2003. The genetic legacy of the Mongols. Am J Hum Genet 72:717–721.

Zhivotovsky LA, Underhill PA, Cinnioglu C, Kayser M, Morar B, Kivisild T, Scozzari R, Cruciani F, Destro-Bisol G, Spedini G, Chambers GK, Herrera RJ, Yong KK, Gresham D, Tournev I, Feldman MW, Kalaydjieva L. 2004. The effective mutation rate at Y chromosome short tandem repeats, with application to human population-divergence time. Am J Hum Genet 74:50–61.