Short Report

The Y-Chromosome Short Tandem Repeats Variation Within Haplogroup E3b: Evidence of Recurrent Mutation in SNP

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ABSTRACT Haplogroup E3b is defined by a single nucleotide mutation (SNP) in locus M35 and is found at high frequency (more than 35%) in populations from North Africa with a heterogeneous distribution. On the basis of compilation of 553 Y-chromosomes from Europe and 633 from sub-Saharan Africa we selected 130 individuals belonging to haplogroup E3b and characterized subhaplogroups according to the Y-Chromosome Consortium nomenclature. Y-chromosome haplotypes can be defined using short tandem repeats (STR). The use of STRs makes it possible to measure diversity and estimate age coalescence. Significant differences on frequencies of Y-chromosome STR loci were found among the E3b subhaplogroups and the same was observed when haplotype frequencies were considered. Some mutations in SNPs were detected when comparing E3b subhaplogroups with the correspondent STR haplotypes. These results show that the mutation rate for some SNPs could be higher than previously thought and also that it is important to associate both haplotype and haplogroup in Y-chromosome studies. Am. J. Hum. Biol. 20:185–190, 2008.

The human Y chromosome haplogroup E3b is characterized by mutations YAP, SRY4064, and M35 according to the phylogeny and haplogroup nomenclature proposed by the Y-Chromosome Consortium (Jobling and Tyler-Smith, 2003; YCC, 2002). The haplogroup E3b is frequent in northern Africa and also in Europe and western Asia (Semino et al., 2004) because of migrations of farmers at the time of the Neolithic expansion (Underhill et al., 2001). Parallel to the paragroup M35 (E3b*) found in east and south of Africa (Semino et al., 2004), the three main subclades of haplogroup E3b can be defined by SNP mutations E-M78 (E3b1), E-M81 (E3b2), and E-M123 (E3b3) (Jobling and Tyler-Smith, 2003; YCC, 2002). None of these clades are homogeneously distributed throughout the African continent. E3b1 is the most common subhaplogroup, with a distribution in north and east of Africa, E3b2 appears in north of Africa associated to Berber-speaking populations, and E3b3 was more likely introduced in Ethiopia from the Near East and is today mainly present in east of Africa (Semino et al., 2004).

Haplogroups are considered to be stable because of the very low mutation rate of most binary markers (SNPs), around 10^{-9} per base per generation (Thomson et al., 2000), showing evidence of recurrent mutation at only 6 of 240 SNPs (YCC, 2002). On the contrary, the mutation rate in short tandem repeats (STRs) is higher (just about 10^{-4}) and varies with the mean number of repeats and the motif length (Forster et al., 2000). Nevertheless, mutations in some SNPs were described as being associated to gene conversion events (Rozen et al., 2003).

The aim of the present work is to define a Y-chromosome STR profile within each E3b subhaplogroup, to observe if there are haplotype differences within the haplogroups and to find possible mutations in SNPs.

MATERIALS AND METHODS

DNA samples

We have selected 130 E3b chromosomes from a compilation of 1,186 unrelated men already typed for Y-chromosome SNPs, from European populations of mainland Portugal north ($N = 101$), center ($N = 102$) and south ($N = 100$), Madeira ($N = 129$) and Azores ($N = 121$) archipelagos (Gonçalves et al., 2005), and African populations from Cabo Verde north ($N = 101$) and south ($N = 100$), Guinea-Bissau ($N = 282$) (Gonçalves et al., 2003), and São Tomé e Príncipe ($N = 150$) (Gonçalves et al., 2007).

Binary markers typing

The binary markers M35, M78, M81, and M123 were typed according to Underhill et al. (2001) and the classification of E3b subhaplogroups was done according to Y-chromosome Consortium (Jobling and Tyler-Smith, 2003; YCC, 2002).

Y-STR typing

To further define the variation and the coalescence time of each E3b subhaplogroup, nine STR loci (DYS19, DYS389 I, DYS389 II, DYS390, DYS391, DYS392, DYS393, DYS438, and DYS439 from Powerplex Y system kit (Promega Corporation, Madison, WI) were assayed using standard methodology.

The allele 9 of STR DYS460 (or GATA7.1) was tested to further define E3b1 cluster α and allele 10 of STR DYS439 was used to define E3b1 cluster β whereas, allele 11 from STR DYS19 was used to define E3b1 cluster γ (Cruciani et al., 2004). The samples from E3b1 subhaplogroup that presented none of these alleles were designated as E3b1,* because there seems to be an assemblage of all those who are not included in cluster α, cluster β, or cluster γ and could represent a whole of other clusters not yet defined.

Data analysis

The coalescence time of STR variation was estimated as the average squared difference in the number of repeats

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TABLE 1. Y-chromosome haplotypes defined by 11 STR loci within E3b sub haplogroups for the populations of Azores (AZ, n = *11), Madeira (MD,* n = *18), Portugal north (NP,* n = *21), Portugal center (CP,* n = *15), Portugal south (SP,* n = *8), Cabo Verde north (CVN,* n = *27), Cabo Verde south (CVS,* n = *13), and Guinea-Bissau (GN,* n = *17)*

Hg		DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385	DYS438	DYS439	DYS460	AZ	MD	NP	CP	SP	CVN	CVS	GN
E3b*	H1	13	12	30	22	9	11	13	12,15	10	13									1
E3b*	H2	13	13	29	24	11	11	14	14,16	10	13									1
E3b*	H3	13	13	30	22	9	11	12	13,16	10	13									1
E3b*	H4	13	13	30	22	9	12	12	16,17	10	12									1
E3b*	H5	13	13	30	23	9,10	11	13	12,15	10	12,13			2						
E3b*	H6	13	13	30	23	10	13	13	16,16	11	12									1
E3b*	H7	13	13	30	24	10	10	13	15,16	10	13									1
E3b*	H8	13	13	30	24	10	11	12	16,17	10	13							1		
E3b*	H9	13	13	31	24	10	11	13	16,18	11	12						1			
E3b*	H10	13	14	30	24	9	11	13	13,14	9	10				1					
E3b*	H11	13	14	30	24	9	11	13	13,14	11	10				2					
E3b*	H12	13	14	31	24	10	11	13	16,17	10	12				1					
E3b*	H13	13	14	31	24	10	11	12	16,19	11	12				1					
E3b*	H14	16	12	29	22	10	11	13	15,16	8	11							1		
E3b1α	H15	13	13	29	24	9	11	13	17,18	10	11	9		1						
E3b1α	H16	13	13	29	24	10	11	13	16,17	10	11	9		1						
E3b1α	H17	13	13	29	24	10	11	13	16,18	10	11	9		2						
E3b1α	H18	13	13	29	24	10	11	13	17,18	10	11	9		1						
E3b1α	H19	13	13	30	23	10	11	13	17,17	10	11	9							1	
E3b1α	H20	13	13	30	23	10	11	13	16,17	10	11	9							1	
E3b1α	H21	13	13	30	23	10	11	13	17,18	10	11	9			1					
E3b1α	H22	13	13	30	24	10	11	12	16,19	10	12	9				2				
E3b1α	H23	13	13	30	24	10	11	13	16,18	10	12	9			1					
E3b1α	H24	13	13	30	24	10	11	13	17,19	10	12	9							1	
E3b1α	H25	13	13	31	23	11	11	13	16,17	10	12	9					1			
E3b1α	H26	13	13	31	24	10	11	13	16,16	10	11	9	1							
E3b1α	H27	13	14	31	25	10	11	13	16,17	10	10	9			1					
E3b1β	H28	13	12	29	23	10	11	13	17,18	10	10	10			1					
E3b1β	H29	13	12	29	24	10	11	13	17,18	10	10	10							1	
E3b1β	H30	13	12	29	24	10	11	13	17,18	10	10	11						3	3	
E3b1β	H31	13	13	31	24	11	11	13	15,16	10	10	10								1
E3b1β	H32	14	13	30	23	10	11	14	13,14	10	10	10								1
E3b1*	H33	13	13	30	23	9,10	11	13	12,15	10	12,13	10		1						
E3b1*	H34	13	13	30	23	10	11	13	16,16	10	11	11	1							
E3b1*	H35	13	13	30	23	10	11	13	17,17	10	11	10						1		
E3b1*	H36	13	13	30	23	10	11	13	16,18	10	12	10		1						
E3b1*	H37	13	13	30	23	10	13	13	16,16	11	12	11						1		
E3b1*	H38	13	13	30	23	11	11	14	16,17	10	12	11	1							
E3b1*	H39	13	13	30	24	10	11	13	17,17	10	12	12								1
E3b1*	H40	13	13	30	24	10	11	13	17,17	10	13	10			1					
E3b1*	H41	13	13	30	24	10	11	13	16,17	10	12	12								2
E3b1*	H42	13	13	30	24	10	11	13	16,18	10	12	10				1		1		
E3b1*	H43	13	13	30	24	10	11	14	16,16	10	13	11								1
E3b1*	H44	13	13	30	24	10	12	13	15,15	10	12	11								1
E3b1*	H45	13	13	30	24	11	11	13	16,18	10	12	10						2		
E3b1*	H46	13	13	30	24	11	11	13	17,18	10	12	11						4		
E3b1*	H47	13	13	30	24	11	11	14	16,18	10	13	10				1				
E3b1*	H48	13	13	30	25	10	11	13	15,17	10	12	11				1				
E3b1*	H49	13	13	30	25	11	11	13	17,18	10	11	10	1							
E3b1*	H50	13	13	31	22	10	11	13	14,16	10	11	12						1		
E3b1*	H51	13	13	31	23	10	11	13	15,16	10	11	11								1
E3b1*	H52	13	13	31	24	10	12	12	15,15	10	12	11								1
E3b1*	H53	13	13	31	24	10	12	14	16,16	10	13	12								1

(Continued)

TABLE 1. (Continued)

Hg		DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385	DYS438	DYS439	DYS460	AZ	MD	NP	CP	SP	CVN	CVS	GN
E3b1*	H54	13	13	32	25	11	11	13	15,18	10	12	11								1
E3b1*	H55	13	14	31	23	10	11	13	16,16	11	12	11						1		
E3b1*	H56	13	14	31	25	10	11	14	14,18	10	13	10				1				
E3b1*	H57	14	12	29	24	10	12	12	17,17	10	11	11			1					
E3b1*	H58	14	13	30	25	10	11	13	17,20	10	12	10			1					
E3b1*	H59	14	13	31	25	10	11	13	16,17	10	11	11			1					
E3b2	H60	13	13	29	24	9	11	13	13,13	10	11			2						
E3b2	H61	13	13	29	24	9	11	13	13,14	9	10				1					
E3b2	H62	13	13	29	24	9	11	13	13,14	10	10		1		2				1	
E3b2	H63	13	13	29	24	10	11	13	13,14	10	9					1				
E3b2	H64	13	13	29	25	9	11	13	13,14	10	10				1					
E3b2	H65	13	13	30	24	11	11	13	13,14	12	11		1							
E3b2	H66	13	13	30	25	11	11	13	17,18	10	11		1							
E3b2	H67	13	13	31	24	10	12	13	15,16	10	12								1	
E3b2	H68	13	14	30	23	9	11	13	13,14	9	10						1			
E3b2	H69	13	14	30	23	9	11	13	13,14	10	10			1			1			
E3b2	H70	13	14	30	24	9	11	12	13,14	10	10		1							
E3b2	H71	13	14	30	24	9	11	13	13,13	10	10							1		
E3b2	H72	13	14	30	24	9	11	13	14,14	10	10					1		1		
E3b2	H73	13	14	30	24	9	11	13	14,14	10	11		1							
E3b2	H74	13	14	30	24	9	11	13	13,14	10	9						1			
E3b2	H75	13	14	30	24	9	11	13	13,14	10	10			1	2	3	1		1	
E3b2	H76	13	14	30	24	9	11	13	13,14	10	12								1	
E3b2	H77	13	14	30	24	9	11	13	13,15	10	10			1						
E3b2	H78	13	14	30	24	9	11	14	13,14	10	10		1							
E3b2	H79	13	14	30	24	11	11	14	13,14	10	11						1			
E3b2	H80	13	15	31	24	9	11	13	14,14	10	10								1	
E3b2	H81	14	13	30	23	9	11	13	14,14	10	10			1						
E3b2	H82	14	14	30	23	9	11	13	13,14	10	10			1						
E3b2	H83	14	14	31	22	9	11	12	13,14	10	10						1			
E3b3	H84	13	12	29	23	10	11	12	15,16	10	12					1				
E3b3	H85	13	12	30	24	10	11	13	16,16	10	13				1					
E3b3	H86	13	12	30	24	10	11	13	16,17	10	12					1				
E3b3	H87	13	13	31	24	10	11	13	17,19	10	11							1		
E3b3	H88	13	13	31	24	10	11	14	16,16	10	12		1							
E3b3	H89	13	14	30	24	9	11	13	16,17	9	11					1				
E3b3	H90	14	13	31	23	10	11	13	16,18	10	13					1				
E3b3	H91	15	13	30	23	10	11	14	15,17	11	11							1		
E3b3	H92	15	13	30	24	10	11	12	13,18	10	11							1		
E3b3	H93	15	13	30	24	10	11	12	14,18	10	11			1						
E3b3	H94	15	13	31	23	10	11	14	15,17	11	11							4		
E3b3	H95	15	13	31	23	10	11	15	15,17	11	11								1	
E3b3	H96	15	13	31	24	10	11	12	13,18	10	11			1				2		
E3b3	H97	15	14	31	24	10	11	12	13,18	10	10				1					

Haplotype diversity within each sub-haplogroup: E3b*, 0.9833 ± 0.0278; E3b1α, 0.9810 ± 0.0308; E3b1β, 0.5833 ± 0.1833; E3b1*, 0.9822 ± 0.0132; E3b2, 0.9444 ± 0.0266; E3b3, 0.9474 ± 0.0376.

TABLE 2. Allele frequencies and gene diversity (gd) estimated for each STR within E3b sub-haplogroups

	E3b total	E3b1	E3b2	E3b3	E3b*
	$N = 130$	$N = 58$	$N = 37$	$N = 19$	$N = 16$
DYS19					
13	0.838	0.936	0.919	0.316	0.937
14	0.062	0.064	0.081	0.053	
15	0.092			0.631	
16	0.008				0.063
gd	0.287 ± 0.049	0.131 ± 0.058	0.153 ± 0.075	0.526 ± 0.089	0.125 ± 0.106
DYS389I					
12	0.108	0.277		0.158	0.125
13	0.631	0.681	0.351	0.737	0.563
14	0.254	0.042	0.622	0.105	0.313
15	0.008		0.027		
gd	0.530 ± 0.037	0.350 ± 0.071	0.503 ± 0.053	0.444 ± 0.124	0.608 ± 0.090
DYS389II					
29	0.200	0.388	0.243	0.053	0.125
30	0.569	0.431	0.676	0.316	0.688
31	0.223	0.171	0.081	0.632	0.188
32	0.008	0.010			
gd	0.591 ± 0.032	0.612 ± 0.046	0.491 ± 0.076	0.526 ± 0.089	0.508 ± 0.126
DYS390					
22	0.046	0.010	0.027		0.250
23	0.231	0.236	0.135	0.421	0.188
24	0.662	0.671	0.811	0.579	0.563
25	0.062	0.083	0.027		
gd	0.507 ± 0.042	0.551 ± 0.057	0.332 ± 0.092	0.515 ± 0.052	0.625 ± 0.093
DYS391					
9,10	0.023	0.010			0.125
9	0.308	0.022	0.865	0.053	0.375
10	0.546	0.811	0.054	0.947	0.438
11	0.123	0.157	0.081		0.063
gd	0.596 ± 0.029	0.388 ± 0.066	0.249 ± 0.090	0.105 ± 0.092	0.692 ± 0.074
DYS392					
10	0.008				0.063
11	0.931	0.949	0.973	1.000	0.813
12	0.046	0.040	0.027		0.063
13	0.015	0.010			0.063
gd	0.132 ± 0.040	0.163 ± 0.063	0.054 ± 0.051	0.000 ± 0.000	0.350 ± 0.148
DYS393					
12	0.131	0.065	0.054	0.368	0.250
13	0.746	0.852	0.892	0.263	0.688
14	0.115	0.084	0.054	0.316	0.063
15	0.008			0.053	
gd	0.416 ± 0.048	0.305 ± 0.073	0.204 ± 0.086	0.731 ± 0.048	0.492 ± 0.117
DYS438					
8	0.008				0.063
9	0.023		0.027	0.053	0.063
10	0.854	0.958	0.946	0.632	0.563
11	0.100	0.020		0.316	0.313
12	0.015	0.022	0.027		
gd	0.262 ± 0.048	0.101 ± 0.054	0.107 ± 0.068	0.526 ± 0.089	0.617 ± 0.096
DYS439					
9	0.015		0.054		
10	0.323	0.356	0.730	0.053	0.188
11	0.277	0.271	0.162	0.684	0.063
12	0.269	0.313	0.054	0.158	0.313
13	0.092	0.051		0.105	0.313
12,13	0.023	0.010			0.125
gd	0.613 ± 0.073	0.707 ± 0.034	0.447 ± 0.091	0.521 ± 0.123	0.800 ± 0.057
DYS385					
13,13	0.023		0.081		
14,14	0.038		0.135		
15,15	0.015	0.020			
16,16	0.069	0.073		0.105	0.063
17,17	0.038	0.063			
12,15	0.031	0.010			0.188
13,14	0.231		0.703		0.188
13,15	0.008		0.027		
13,16	0.008				0.063
13,18	0.038			0.263	
14,16	0.015	0.010			0.063
14,18	0.015	0.010		0.053	
15,16	0.046	0.043	0.027	0.053	0.125

(Continued)

TABLE 2. (Continued)

	E3b total	E3b1	E3b2	E3b3	E3b*
15,17	0.054	0.010		0.316	
15,18	0.008	0.010			
16,17	0.100	0.129		0.105	0.188
16,18	0.085	0.127		0.053	0.063
16,19	0.023	0.044			0.063
17,18	0.131	0.384	0.027		
17,19	0.015	0.022		0.053	
17,20	0.008	0.010			
gd	0.914 ± 0.012	0.872 ± 0.026	0.519 ± 0.092	0.842 ± 0.057	0.917 ± 0.042

between all current chromosomes and the founder haplotype (defined by the most frequent allele to each STR), divided by $w = 6.9 \times 10^{-4}$ per 25 years (Zhivotovsky et al., 2004). The coalescence time is expressed in kilo (thousand) years ago (kya).

Y-chromosome STR haplotype frequencies were estimated by direct counting. The variability indexes and the comparisons of STR frequencies for each locus within each subhaplogroup were calculated according to Fisher's exact test using the Arlequin software (Schneider et al., 2000). Probability values of $P < 0.05$ were considered as statistically significant.

RESULTS

All haplotypes found harbored within Y-chromosome haplogroups are shown in Table 1. Some individuals share similar haplotypes belonging to different E3b subhaplogroups: for example two E3b* and one E3b1* individuals share a similar haplotype (H5 and H33) characterized by double alleles at DYS391 and DYS438 STR loci. Similar haplotypes (H6 and H37) are shared on an E3b* and E3b1* background, respectively, and the same happens to haplotypes H49 and H66 associated to individuals E3b1* and E3b2.

For instance, DYS391 allele 9 and DYS385 genotype 13,14 appear with a high frequency in the E3b2 subhaplogroup but not in the others where allele sizes are generally higher, varying between 15 and 20 repeats. Moreover, DYS19 allele 15 appears only associated to individuals of the E3b3 subhaplogroup (Table 2). When haplotype frequencies were considered, significant differences ($P < 0.05$) between all the subhaplogroups were found.

Coalescence age of E3b haplogroup is lower (21.1 ± 7.8 kya) than those calculated by Cruciani et al. (2004); 25.6, 95% CI 24.3–27.4 kya but considering the high standard error it could be accepted as in the range. On the contrary, the E3b2 coalescence age of 13.4 ± 5.0 is higher (already considering the standard error) than the one suggested by Cruciani et al. (2004); 5.6, 95% CI 4.6–6.3 kya.

DISCUSSION AND CONCLUSIONS

The existence of three individuals that share a haplotype with a double duplication in two different loci suggests that a recurrent mutation occurred in SNP M78 because the duplication event is rare (Butler et al., 2005) and it is unlikely to occur twice. For the two individuals sharing the same haplotype (H49 and H66) but belonging to different E3b subhaplogroups (E3b1* and E3b2), two situations may have occurred: two recurrent mutations in M78 and M81, or several STR mutations including one in DYS385 that changes from a probable 13,14 that is associated to 70% of individuals belonging to E3b2 haplogroup to an 17,18 that appears in 38% of individuals from E3b1 haplogroup (Table 2).

In support of previous examples, one should be careful when suggesting the creation of new branches and new classifications like in Cruciani et al. (2004), where a new organization of E3b subhaplogroups is presented based only on two individuals mutation (<1%). Taking in consideration the cases described in this article, it is possible that the derivate state at M215 or the ancestral state at M35 occur because of a recurrent mutation in M35 or a recombination in case of M215 that is located in the X-homolog region. Therefore, we suggest using only the Y-Chromosome Consortium nomenclature, avoiding the new nomenclature proposed by Cruciani et al. (2004). Nevertheless, the clusters defined by Cruciani et al. (2004) within subhaplogroup E3b1 can be a useful tool to distinguish the various geographic influences in populations but may sometimes generate some confusion because the mutation rate of DYS439 is the highest of the STR set used (Gusmão et al., 2005). For example, haplotype H27 belongs to an individual that had the allele 10 from DYS439 and also the allele 9 from DYS460 and can therefore be considered as belonging to cluster α or cluster β. Likewise, H35 haplotype is similar to haplotype H19 (E3b1α) but is classified as E3b1*, because it did not harbor allele 9 from DYS460. These examples show how careful one must be when using STR data to define a particular cluster, especially if the mutation rate of the STR is high, as with DYS460 and DYS439 (Gusmão et al., 2005).

The fact that some individuals share the same haplotype and belong to different E3b subhaplogroups suggests that SNP mutations may occur frequently, like the recurrent mutations in M78 that seem to occur four times, probably because of mechanisms of gene conversion.

The data presented here suggests that using SNPs and STRs information together provides a more reliable characterization of an individual in terms of Y-chromosome origin, minimizing the risk of misassignment because of mutations.

LITERATURE CITED

Butler JM, Decker AE, Kline MC, Vallone PM. 2005. Chromosomal duplications along the Y-chromosome and their potential impact on Y-STR interpretation. J Forensic Sci 50:853–859.

Cruciani F, La Fratta R, Santolamazza P, Sellito D, Pascone R, Moral P, Warson E, Guida V, Colomb EB, Zaharova B, Lavinha J, Vona G, Aman R, Cali F, Akar N, Richards M, Torroni A, Novelleto A, Scozzari R. 2004. Phylogeographic analysis of haplogroup E3b (E-M215) Y chromosomes reveals multiple migratory events within and out of Africa. Am J Hum Genet 74:1014–1022.

Forster P, Rohl A, Lunnemann P, Brinkmann C, Zerjal T, Tyler-Smith C, Brinkmann B. 2000. A short tandem repeat-based phylogeny for the human Y chromosome. Am J Hum Genet 67:182–196.

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 Açores record elements of Sephardim and Berber ancestry. Ann Hum Genet 69:443–454.

Gonçalves R, Rosa A, Freitas A, Fernandes A, Kivisild T, Villems R, Brehm A. 2003. Y-chromosome lineages in Cabo Verde Islands witness the diverse geographic origin of its first male settlers. Hum Genet 113:467–472.

Gonçalves R, Spínola, Brehm A. 2007. Y-chromosome lineages in São Tomé e Príncipe islands: evidence of European influence. Am J Hum Biol 14:422–428.

Gusmão L, Sanchez-Diz P, Calafell F, Martin P, Alonso CA, Alvarez-Fernandez F, Alves C, Borjas-Fajardo L, Bozzo WR, Bravo ML, Builes JJ, Capilla J, Carvalho M, Castillo C, Catanesi CI, Corach D, Di Lonardo AM, Espinheira R, Fagundes de Carvalho E, Farfan MJ, Figueiredo HP, Gomes I, Lojo MM, Marino M, Pinheiro MF, Pontes ML, Prieto V, Ramos-Luis E, Riancho JA, Souza Goes AC, Santapa OA, Sumita DR, Vallejo G, Vidal Rioja L, Vide MC, Vieira da Silva CI, Whittle MR, Zabala W, Zarrabeitia MT, Alonso A, Carracedo A, Amorim A. 2005. Mutation rates at Y chromosome specific microsatellites. Hum Mutat 26:520–528.

Jobling MA, Tyler-Smith C. 2003. The human Y chromosome: An evolutionary marker comes of age. Nat Rev Genet 4:598–612.

Schneider S, Kueffer JM, Roessli D, Excoffier L. 2000. Arlequin: a software for population genetic data analysis. Switzerland: Genetics and Biometry Laboratory, University of Geneva.

Semino O, Magri C, Benuzzi G, Lin AA, Al-Zahery N, Battaglia V, Maccioni L, Triantaphyllidis C, Shen P, Oefner PJ. 2004. Origin, diffusion and differentiation of Y chromosome haplogroups E and J. Inferences on the neolithization of Europe and later migratory events in the Mediterranean area. Am J Hum Genet 74:1023–1034.

Rozen S, Skalettsky H, Marszalek JD, Minx PJ, Cordum HS, Waterston RH, Wilson RK, Page D. 2003. Abundant gene conversion between arms of palindromes in human and ape Y chromosomes. Nature 423:873–876.

Thomson R, Pritchard JK, Shen P, Oefner PJ, Feldman MW. 2000. Recent common ancestry of human Y chromosomes: evidence from DNA sequence data. Proc Natl Acad Sci USA 97:7360–7365.

Underhill P, 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.

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

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, Tounev 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.