Legal Medicine 14 (2012) 105–109

Contents lists available at SciVerse ScienceDirect

Legal Medicine

journal homepage: www.elsevier.com/locate/legalmed

Announcement of Population Data

Y chromosome STR allelic and haplotype diversity in a Rwanda population from East Central Africa

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ARTICLE INFO

Article history:
Received 23 August 2011
Received in revised form 4 November 2011
Accepted 8 December 2011

Keywords:
Y-STR
Rwanda population
African population
Y-STR haplotypes
Gene diversity
Genetic distance

ABSTRACT

We have analyzed 17 Y-chromosomal STR loci in a population sample of 69 unrelated male individuals of the Rwanda-Hutu population from East Central Africa using an AmpFlSTR® Yfiler™ PCR amplification kit. A total of 62 unique haplotypes were identified among the 69 individuals studied. The haplotype diversity was found to be 0.9970 for this population. The gene diversity ranged from 0.1130 (DYS392) to 0.7722 (DYS385). Comparison of populations in this study with twenty-five other national and global populations using Principal Co-ordinate Analysis (PCA) and phylogenetic molecular analysis using a genetic distance matrix indicates a delineation of all the African populations from other unrelated populations. The results of population pair-wise Fst *p* values indicate statistically significant differentiation of the Rwandan population when compared with 25 other global populations including four African populations ($p = 0.0000$). Analysis of Molecular Variance (AMOVA) of the Rwanda population with four other African populations indicated a 93% variance within populations and 7% variance among the five populations. A data base search of the 62 haplotypes yielded only one non-African haplotype match, suggesting these haplotypes are unique to the African continent.

Populations: A total of 69 unrelated male samples (out of 100 samples collected) from the Republic of Rwanda located in East Central Africa were analyzed for seventeen Y chromosomal STR loci.

Extraction: Blood samples were collected with informed consent from all individuals. Samples were collected in accordance with the ethical guidelines stipulated by the institutions involved in this study. DNA was isolated following the method as described by Budowle et al. [1]. Total human DNA was quantitated using a Quantifiler® Human DNA quantitation kit per manufacturer's recommendations (Applied Biosystems, Foster City, CA).

PCR: Approximately one nanogram of DNA template was amplified at 17 Y chromosomal STR loci in a multiplex reaction using the AmpFlSTR® Yfiler™ PCR amplification kit (Applied Biosystems, Foster City, CA). The seventeen Y-STR loci amplified are DYS19, DYS385a, DYS385b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 and YGATAH4.

Typing: Amplicons were separated by capillary electrophoresis on an automated ABI 310 Genetic Analyzer (Applied Biosystems, Foster City, CA) and sized with a Genescan-500 LIZ size standard. Allele calls and genotyping were carried out using the allelic ladder provided in the kit and GeneMapper® ID v3.2 software (Applied Biosystems, Foster City, CA).

Quality control: Positive and negative controls as specified in the Yfiler™ kit user's manual. The data were analyzed and verified by two independent analysts.

Analyses of data: STR allele frequencies and gene diversity estimates were calculated using Genepop version 4.0 [2,3]. The haplotype diversity was estimated using Nei's formula $h = [n(1 - \sum x_i^2)]/(n - 1)$, where n is the number of samples analyzed and x_i is the frequency of the *ith* haplotype [4]. The frequency of the Y STR haplotypes and population pair-wise Fst *p* values were calculated using Arlequin v 3.1 [5]. Nei's genetic distances of the samples from the current study as well as 25 other national and global populations were calculated from the allele frequencies of respective populations using POPTREE2 for phylogenetic and molecular evolutionary analysis [6]. Graphical representation of genetic distances of the population in this study along with other national and global populations was performed based on Principal Co-ordinate Analysis (PCA) plot using GenAlEx software package version 6.3 [7]. Analysis of Molecular Variance (AMOVA) was also performed by the same software package.

Results: A total of 62 unique haplotypes were identified among the 69 individuals studied, with seven shared haplotypes, each shared by two individuals. The unique haplotypes observed in this study along with their frequency are presented in Table 1. Allele

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doi:10.1016/j.legalmed.2011.12.002

Table 1
The unique Y STR haplotypes and their frequency in the Rwanda population.

Haplotype	Sample	DYS19	DYS 385	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 437	DYS 438	DYS 439	DYS 448	DYS 456	DYS 458	DYS 635	Y_GATA_H4	Frequency
1	RW-1	16	17, 18	13	30	21	10	11	14	14	11	12	20	16	16	19	11	1
2	RW-2	16	16, 19	13	30	21	10	11	15	14	11	11	21	17	17	21	10	1
3	RW-3	15	17, 21	13	30	21	10	11	15	14	11	12	21	16	16	21	11	1
4	RW-4, 61	15	16, 18	13	30	21	10	11	13	14	12	11	20	15	16	21	12	2
5	RW-5	17	16, 19	14	31	21	10	11	15	15	11	12	21	18	17	22	11	1
6	RW-6	15	16, 16	13	30	21	10	11	14	14	11	12	21	16	18	24	11	1
7	RW-9	15	15, 17	13	31	21	10	11	13	14	12	12	21	15	18	21	12	1
8	RW-11	16	14, 19	13	29	21	10	11	14	14	11	13	21	16	16	21	12	1
9	RW-12	15	15, 18	13	30	21	10	11	14	14	11	12	21	15	16	22	12	1
10	RW-13, 82	16	17, 19	14	31	21	10	11	15	14	11	12	21	16	16	21	11	2
11	RW-14	16	17, 20	13	30	21	10	11	15	14	11	13	21	16	15	21	11	1
12	RW-15	16	15, 16	13	30	21	10	11	13	14	11	11	23	15	17	23	12	1
13	RW-16, 88	15	15, 15	12	29	24	11	11	13	15	12	12	19	16	18	18	12	2
14	RW-17	15	15, 18	13	30	21	10	11	14	14	11	12	21	15	16	21	12	1
15	RW-18	14	14, 19	12	28	27	10	11	13	14	11	11	19	15	18	24	11	1
16	RW-22, 25	16	17, 19	13	32	21	10	11	14	14	11	13	20	16	16	21	11	2
17	RW-23	15	16, 17	13	30	21	10	11	14	14	11	12	21	16	18	20	11	1
18	RW-24	15	17, 19	14	31	21	10	11	14	14	11	21	17	1821	18	21	11	1
19	RW-26	15	17, 19	14	31	21	10	11	15	14	11	12	21	17	17	21	11	1
20	RW-27, 66	16	17, 17	13	30	21	9	11	14	14	11	12	21	16	16	21	12	2
21	RW-28	16	16, 18	14	31	21	10	11	14	14	12	12	22	16	16	22	11	1
22	RW-30, 77	15	17, 19	14	31	21	10	11	15	14	11	11	21	17	17	21	11	2
23	RW-32	15	11, 12	13	31	24	10	12	14	15	10	12	20	13	18	17	11	1
24	RW-34	17	18, 19	13	30	20	10	11	15	14	11	13	21	17	16	21	11	1
25	RW-38	15	16, 18	14	31	21	10	12	16	14	11	12	21	16	17	21	11	1
26	RW-39	15	15, 15	13	30	21	11	11	14	14	11	12	21	15	16	21	13	1
27	RW-40	17	15, 18	13	30	21	21	10	11	14	14	11	11	15	15	21	11	1
28	RW-41, 71	13	16, 16	14	31	23	11	11	13	14	10	11	20	16	16	21	11	2
29	RW-43	15	15, 18	14	31	21	10	11	14	14	11	12	21	15	16	21	12	1
30	RW-45	15	15, 15	12	30	24	10	11	13	15	11	13	19	15	17	21	12	1
31	RW-46	16	17, 17	13	30	21	10	11	15	14	10	13	20	17	16	20	10	1
32	RW-48	16	12, 12	12	30	24	10	11	14	15	11	11	21	12	19	18	11	1
33	RW-49	17	17, 17	13	30	21	11	11	13	14	11	11	21	15	16	21	11	1
34	RW-50	15	16, 17	13	30	21	10	11	14	14	11	12	21	15	19	22	11	1
35	RW-51	16	16, 18	14	31	21	10	11	14	14	11	12	21	16	18	21	12	1
36	RW-52	14	14, 19	12	28	25	10	11	13	14	11	11	19	15	17	24	11	1
37	RW-53	15	16, 17	13	31	21	10	11	13	14	11	11	21	15	16	21	11	1
38	RW-54	17	17, 18	13	30	21	10	11	14	14	11	11	21	15	16	21	11	1
39	RW-55	15	17, 20	14	32	22	10	11	14	14	11	12	21	15	16	21	11	1
40	RW-56	17	17, 19	14	31	21	10	11	15	14	11	11	21	16	18	22	11	1
41	RW-57	16	16, 17	13	30	21	10	11	14	14	11	11	21	16	18	22	11	1
42	RW-59	15	13, 17	14	30	24	10	11	13	14	11	11	19	14	15	23	9	1
43	RW-60	16	17,17	13	31	21	10	11	16	14	11	11	20	16	17	21	11	1
44	RW-63	16	17,19	13	30	21	10	11	14	14	11	11	21	16	16		21	11
45	RW-67	14	14, 19	12	29	24	10	11	13	14	11	11	19	15	18	23	11	1
46	RW-69	15	17, 20	13	33	21	10	11	14	14	11	13	20	16	16	21	11	1
47	RW-70	17	19, 19	14	31	21	10	11	15	14	11	12	21	17	17	22	11	1
48	RW-72	15	16, 18	13	31	21	10	11	14	14	10	13	22	16	16	21	12	1
49	RW-73	15	16,17	13	30	21	10	11	14	14	11	12	21	15	18	22	11	1
50	RW-79	15	16,18	13	32	21	10	11	13	14	12	11	21	15	17	21	12	1
51	RW-81	15	17, 18	13	30	21	10	10	14	14	11	12	21	16	17	22	11	1
52	RW-83	15	15,17	13	30	21	10	11	14	14	12	13	22	15	16	22	12	1
53	RW-84	17	17, 19	14	30	21	10	11	15	14	11	12	21	17	17	21	11	1
54	RW-85	17	17, 19	14	31	21	10	11	16	14	11	13	21	17	18	22	11	1
55	RW-86	16	17, 17	13	30	21	10	11	15	14	11	12	21	15	17	21	11	1
56	RW-87	15	16, 17	13	31	21	11	11	13	14	12	12	21	16	17	21	12	1
57	RW-90	15	13, 15	14	30	24	11	13	13	14	12	13	19	15	18	23	12	1
58	RW-91	16	17,18	13	31	21	10	11	14	14	11	12	20	15	16	19	11	1
59	RW-92	17	18, 20	14	31	21	10	11	14	14	11	13	22	17	17	21	11	1
60	RW-93	15	16,17	14	31	21	10	11	14	15	11	12	21	15	20	22	11	1
61	RW-95	15	15, 18	14	31	21	11	11	14	14	11	12	21	15	17	22	12	1
62	RW-100	17	17, 19	14	31	21	10	11	15	14	11	12	21	17	17	22	11	1

frequencies, gene diversity and haplotype diversity are presented in Table 2 (supplementary material). The lowest gene diversity (0.1130) was observed at locus DYS392 while the highest gene diversity (0.7722) was observed at locus DYS385. Similar observations of low and high gene diversities for loci DYS392 and DYS385, respectively have been reported in other African populations from Namibia, South Africa and Equatorial Guinea [8–10]. The haplotype diversity was found to be 0.9970 for this population.

Haplotype database comparisons: All the 62 haplotypes with 17 loci data were searched against the YHRD database that contained the same 17 loci data for 31, 860 haplotypes (http://www.yhrd.org) with no matching results indicating the uniqueness of the haplotypes in this group of individuals. A haplotype search against the Applied Biosystems Y STR database (http://www6.appliedbiosystems.com/yfilerdatabase) generated 1 match, with a sample from a Hispanic population. The frequency of the