



Y-chromosome STRs DYS385, DYS19, DYS389-I and II, DYS390, DYS391, DYS392 and DYS393 in five African populations

V. Lopes^a, M. Carvalho^a, S. Antunes^a, M.J. Anjos^a,
L. Andrade^a, M.V. Santos^b, F. Corte-Real^{a,b}, D.N. Vieira^{a,b},
J.J. Gamero^c, M.C. Vide^{a,*}

^a*Instituto Nacional de Medicina Legal, Delegação de Coimbra, Serviço de Genética Forense,
Largo da Sé Nova, 3000-213 Coimbra, Portugal*

^b*Faculty of Medicine, University of Coimbra, Coimbra, Portugal*

^c*Department of Legal Medicine, Faculty of Medicine, University of Cádiz, Cádiz, Spain*

Abstract

Background: The Y chromosome has been used to compare the relationship between populations, representing a rich source of potential information to trace paternal lineages and providing a record of our relatedness. Among different population groups, African populations seem to be very interesting to study, considering the theory of the origin of modern humans and the ethnic variability usually existing. *Methods:* Five male populations from Angola ($n=48$), Cap Verde ($n=47$), Guinea-Bissau ($n=32$), Mozambique ($n=36$) and S. Tome and Prince ($n=30$) were studied for Y-chromosome Short Tandem Repeats (STRs) DYS385, in addition to DYS19, DYS389-I and II, DYS390, DYS391, DYS392 and DYS393 previously searched. *Results:* The high DYS385 polymorphism confers a considerable variability to the Y-chromosome haplotype. *Conclusions:* The comparisons between the African populations and other data, namely from a Portuguese population, showed an interesting location of insular populations, with an intermediate position between Caucasoid and African continental populations, probably due to the European presence in the colonization of those islands.

© 2003 Elsevier Science B.V. All rights reserved.

Keywords: Y-chromosome haplotype; Y-chromosome STRs; African populations; Population genetics

* Corresponding author. Tel.: +351-239-854-230; fax: +351-239-820-549.

E-mail address: mcvide@ci.uc.pt (M.C. Vide).

1. Introduction

The aim of this work is to extend the study of Y-chromosome haplotypes of five African populations to the STR DYS385 in addition to the DYS19, DYS389-I and II, DYS390, DYS391, DYS392 and DYS393 previously searched [1] in order to point out their relative position along with a Caucasian population (Central Portugal) [2].

2. Material and methods

DNA was extracted from air-dried bloodstains from unrelated males, using the Chelex 100 extraction method [3]. Amplification conditions: singleplex DYS385 according to Schneider et al. [4]; multiplex DYS19/DYS389 I and II/DYS390/DYS393 according to Gusmão et al. [5]; multiplex DYS391/DYS392/DYS393 as described by Kloosterman et al. [6]. The detection was carried on ABI 377 DNA sequencers.

The haplotype diversity and the correspondent standard error were calculated according to Nei [7] ($1 - \sum f_i^2$ where f_i is the relative frequency of each haplotype in the population). The mean number of pairwise differences between all pairs of haplotypes for one population was performed according to Tajima ($\sum \sum p_i p_j d_{ij}$; $j < i$ where p_i is the frequency of the i th haplotype and d_{ij} is an estimate of the number of mutations occurred since the divergence of haplotypes i and j) [8].

The probability of obtaining different haplotypes when sampling two individuals from two different populations (differentiation probability) was calculated according to Nei [7]

Table 1

Haplotype diversity (Hapl. Div.) and correspondent standard error, and the mean number of pairwise differences calculated for the five African populations

| Population | <i>N</i> | Nb. Hapl. | Hapl. Div. (%) | Mean Nb. pairwise differences |
|--------------------|----------|-----------|----------------|-------------------------------|
| Angola | 48 | 42 | 97.40 ± 0.18 | 4.24 |
| Cap Verde | 47 | 40 | 97.06 ± 0.31 | 5.69 |
| Guinea-Bissau | 32 | 29 | 96.09 ± 0.48 | 3.59 |
| Mozambique | 36 | 34 | 96.91 ± 0.21 | 5.63 |
| S. Tome and Prince | 30 | 29 | 96.44 ± 0.21 | 5.41 |

N is the total of individuals and Nb. Hapl. is the number of different haplotypes found in each population.

Table 2

Shared haplotypes between all the six populations

| | Angola | Cap Verde | Guinea-Bissau | Mozambique | S. Tome and Prince | Portugal (central) |
|--------------------|--------|-----------|---------------|------------|--------------------|--------------------|
| Angola | – | 1 | 1 | 4 | 2 | 0 |
| Cap Verde | 1 | – | 0 | 0 | 2 | 3 |
| Guinea-Bissau | 1 | 0 | – | 0 | 0 | 0 |
| Mozambique | 4 | 0 | 0 | – | 0 | 0 |
| S. Tome and Prince | 2 | 2 | 0 | 0 | – | 2 |
| Portugal (central) | 0 | 3 | 0 | 0 | 2 | – |

Table 3
Matrix of differentiation probability

| | Angola | Cap Verde | Guinea-Bissau | Mozambique | S. Tome and Prince | Portugal (central) |
|--------------------|--------|-----------|---------------|------------|--------------------|--------------------|
| Angola | – | 0.9991 | 0.9987 | 0.9971 | 0.9979 | 1.0000 |
| Cap Verde | 0.9991 | – | 1.0000 | 1.0000 | 0.9950 | 0.9965 |
| Guinea-Bissau | 0.9987 | 1.0000 | – | 1.0000 | 1.0000 | 1.0000 |
| Mozambique | 0.9971 | 1.0000 | 1.0000 | – | 1.0000 | 1.0000 |
| S. Tome and Prince | 0.9979 | 0.9950 | 1.0000 | 1.0000 | – | 0.9962 |
| Portugal (central) | 1.0000 | 0.9965 | 1.0000 | 1.0000 | 0.9962 | – |

Table 4
Matrix of genetic distances

| | Angola | Cap Verde | Guinea-Bissau | Mozambique | S. Tome and Prince | Portugal (central) |
|--------------------|--------|-----------|---------------|------------|--------------------|--------------------|
| Angola | – | 0.1584 | 0.0793 | 0.1687 | 0.1158 | 0.4485 |
| Cap Verde | 0.1584 | – | 0.1500 | 0.1141 | 0.0421 | 0.1148 |
| Guinea-Bissau | 0.0793 | 0.1500 | – | 0.1343 | 0.1304 | 0.4566 |
| Mozambique | 0.1687 | 0.1141 | 0.1343 | – | 0.1297 | 0.2868 |
| S. Tome and Prince | 0.1158 | 0.0421 | 0.1304 | 0.1297 | – | 0.1451 |
| Portugal (central) | 0.4485 | 0.1148 | 0.4566 | 0.2868 | 0.1451 | – |

$(1 - \sum X_i Y_i)$ where X_i and Y_i are the frequencies of the i th haplotype in X and Y populations, for each shared haplotypes). Nei's genetic distances were determined with Phylip software (version 3.5c) and the corresponding phylogenetic tree was obtained from the software TreeView (version 1.5.2), neighbor-joining method.

3. Results

Table 1 shows the haplotypic diversity (and corresponding standard error) and also the mean number of pairwise differences for each one of the five African populations studied

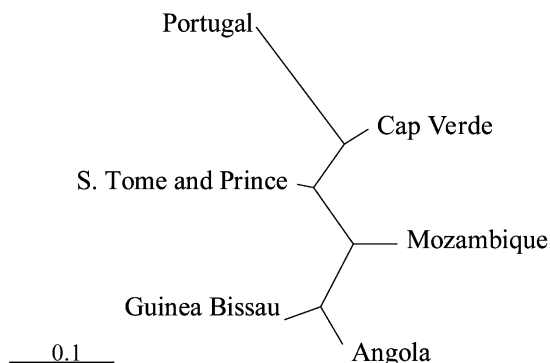


Fig. 1. Phylogenetic tree between the 5 African populations and Central Portugal population.

as well as the number of different haplotypes found. The numbers of shared haplotypes in the six populations are shown in Table 2. Table 3 contains the values of differentiation probability for each pair of populations. These values reflect the values presented in Table 2. The genetic distances calculated from the haplotypic frequencies according to Nei are presented in Table 4 and the corresponding phylogenetic tree is shown in Fig. 1.

4. Discussion

From Table 4 and Fig. 1, we can observe that Angola and Guinea-Bissau are more closely related to each other than with the other African populations as well as Cap Verde and S. Tome and Prince. Mozambique has an intermediate position between those two population groups.

The comparison of Central Portugal along with the five African populations shows that the more closely linked group is the one formed by Cap Verde and S. Tome and Prince, perhaps because their colonisation was made as a result of an admixture between a great number of Portuguese males (and other Caucasoid groups) and African females.

Acknowledgements

The authors would like to thank the staff of Prof. Angel Carracedo (Santiago de Compostela).

References

- [1] F. Corte-Real, M. Carvalho, L. Andrade, M.J. Anjos, C. Pestoni, M.V. Lareu, A. Carracedo, D.N. Vieira, M.C. Vide, Chromosome Y STRs analysis and evolutionary aspects for Portuguese spoken countries, in: G.F. Sensabaugh, P.J. Lincoln, B. Olaisen (Eds.), *Progress in Forensic Genetics*, vol. 8, Elsevier, Amsterdam, 2000, pp. 272–274.
- [2] M. Carvalho, M.J. Anjos, L. Andrade, C. Coxinho, F. Corte-Real, J.J. Gamero, D.N. Vieira, M.C. Vide, Y chromosome polymorphisms: a comparison between Azores and continental Portuguese sample, in: G.F. Sensabaugh, P.J. Lincoln, B. Olaisen (Eds.), *Progress in Forensic Genetics*, vol. 8, Elsevier, Amsterdam, 2000, pp. 302–304.
- [3] P.S. Walsh, D.A. Metzger, R. Higuchi, Chelex 100 as a medium for simple extraction of DNA for PCR-based typing from forensic material, *BioTechniques* 10 (4) (1991) 506–513.
- [4] P.M. Schneider, S. Meuser, W. Waiyawuth, Y. Seo, C. Rittner, Tandem repeat structure of the duplicate Y-chromosomal STR locus DYS385 and frequency studies in the German and three Asian population, *Forensic Sci. Int.* 97 (1998) 61–70.
- [5] L. Gusmão, A. González-Neira, C. Pestoni, M. Brión, M.V. Lareu, A. Carracedo, Robustness of the Y STRs DYS19, DYS389 I and II, DYS390 and DYS393: optimization of a PCR pentaplex, *Forensic Sci. Int.* 106 (1999) 163–172.
- [6] A.D. Kloosterman, M. Pouwels, P. Daselaar, H.J.T. Jansen, Population genetic study of Y-chromosome specific STR loci in Dutch caucasians, in: B. Olaisen, B. Brinkmann, P.J. Lincoln (Eds.), *Progress in Forensic Genetics*, vol. 7, Elsevier, Amsterdam, 1998, pp. 491–493.
- [7] M. Nei, *Molecular Evolutionary Genetics*, Columbia Univ. Press, New York, 1987.
- [8] F. Tajima, Evolutionary relationships of DNA sequences in finite populations, *Genetics* 105 (1983) 437–460.