International Congress Series 1261 (2004) 290–292

www.ics-elsevier.com

Haplotype frequency of the loci DYS437, DYS438, DYS439, DYS460 (Y-GATA A7.1), DYS461 (Y-GATA A7.2), Y-GATA A10, Y-GATA C4 and Y-GATA H4 in the Bahia-Brazil population

M.V. Santos[a,*], M. Carvalho[b], V. Lopes[b], M.C. Vide[b], F. Corte-Real[c], D.N. Vieira[c]

[a] *Faculty of Medicine, University of Coimbra, CNPq/Brazil, Coimbra, Portugal*
[b] *Service of Forensic Genetics, Delegation of Coimbra, National Institute of Legal Medicine, Coimbra, Portugal*
[c] *National Institute of Legal Medicine and Faculty of Medicine, University of Coimbra, Coimbra, Portugal*

Abstract. The study of the Y-chromosome is very important for forensic and population genetics. Although many populations have already been analysed, there is little data about Brazilian populations. The haplotype frequency of eight Y-STRs: DYS437, DYS438, DYS439, DYS460 (Y-GATA-A7.1), DYS461 (Y-GATA-A7.2), Y-GATA-A10, Y-GATA-C4 and Y-GATA-H4 in 72 male individuals from Bahia was established. A total of 67 different haplotypes were observed, of which 63 were unique, three were found in two individuals, and one was found in three individuals. The haplotype diversity and the capacity of discrimination for all eight Y-specific STR loci were calculated to be 98.38% and 93.06%, respectively. A mutational event in both loci DYS437 and DYS439 were found in the same individual. © 2003 Elsevier B.V. All rights reserved.

Keywords: Bahia—Brazil population; Y-STR loci; Population genetic data

---

1. Introduction

The study of the Y-chromosome is used to identify male profiles in crime samples and to determine male lineage and parentage inclusive in the absence of paternal progenitor. The heterogeneity, intra and inter populations, of the Y-STR loci reveal haplogroups

---

* Corresponding author. Delegação de Coimbra, Instituto National de Medicina Legal, Lagro da Sé Nova, 3000-213 Coimbra, Portugal. Tel.: +351-239-814381; fax: +351-239-820549 or Tel./fax: +55-73-616-1809 (Post March, 2003).

E-mail address: diederiche@clix.pt (M.V. Santos).

doi:10.1016/S0531-5131(03)01783-7

representing male ancestral lineage and allow the geographical distribution to be evaluated.

The Bahian population has become very diversified due to the inclusion of a wide range of ethnic backgrounds.

In order to create a database representing the Bahian population, the haplotype frequency of the following eight Y-STRs: DYS437, DYS438, DYS439, DYS460, DYS461, Y-GATA-A10, Y-GATA-C4 and Y-GATA-H4 was established.

2. Materials and methods

The bloodstains of 72 males from Bahia, Brazil were collected and extracted using Chelex procedures [1] and, if necessary, purified by the modified salt–chloroform method.

PCR was performed in accordance with recommendations by the The Spanish and Portuguese ISFG Working Group (GEP-ISFG) for the collaborative study of the Y-Chromosome STRs 2001 [2]. We used two tetraplex systems: GEPY I includes the loci DYS461 (Y-GATA-A7.2), DYS437, DYS438 (with a concentration of 0.72 μM) and Y-GATA-C4; and GEPY II includes DYS460 (Y-GATA-A7.1), Y-GATA-A10, DYS439 and Y-GATA-H4. The primers were those described by Ref. [3] and Ref. [4].

PCR products were separated and detected by capillary electrophoresis on the ABI PRISM® 310 Genetic Analyzer using ROX500 (Applied Biosystems) as Internal Lane DNA Standard, employing ABI software [5] and the GEP-ISFG 2001 exercise reference samples [4] to assign the alleles.

Haplotype frequencies were estimated by gene counting, and haplotype diversity was computed according to Ref. [6] and Ref. [7] using haplotype frequency instead of allele frequency. The capacity of discrimination was also calculated. The study intra and inter populations were done using the Arlequin 2000 [8], PHILIP [9] and Treeview [10] softwares packages.

3. Results and discussion

In the Bahian population, a total of 67 different haplotypes were observed, of which 63 were unique, three were found in two individuals, and one was found in three individuals. The most common (DYS437: 15, DYS438: 12, DYS439: 12, DYS460: 11, DYS461: 12, GATA A10: 15, GATA C4: 23 and GATA H4: 27) had a frequency of 4.2%.

The haplotype diversity and the capacity of discrimination for all eight Y-specific STR loci were calculated to be 98.38% and 93.06%, respectively.

A mutational event in both loci DYS437 and DYS439 were found in the same individual. This sample will be sequenced together with the male descendant sample.

The allele with most frequency were 11 to DYS460, 12 to DYS461, 15 to GATA A10, 23 to GATA C4, 28 to GATA H4, 15 to DYS437, 12 to DYS438 and 12 to DYS439. The DYS439 and GATA C4 loci show the largest gene diversity—70.25% and 68.29%, respectively.

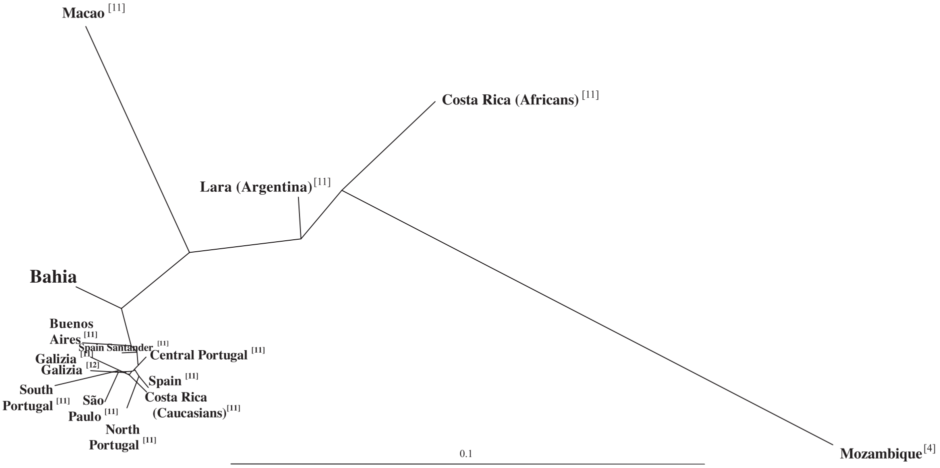

Fig. 1. Genetic tree obtained by method UPGMA using matrix of genetic distances with Φ values. Cited references: [11,12].

A comparison of the results of the pair-wise analysis shown that Bahian population sites in an intermediate position among origin Iberian populations and no Caucasians (Fig. 1).

Acknowledgements

Special thanks go to the donors of the samples. We would like to thank CNPq/Brazil for financial support and "Universidade Estadual de Santa Cruz" and "Departamento de Polícia Técnica—SSP/BA".

References

[1] P.S. Walsh, D.A. Metzger, R. Higuchi, BioTechniques 10 (1991) 506–513.
[2] Available at: http://www.gep-isfg.org.
[3] P. White, et al., Genomics 57 (3) (1999) 433–437.
[4] Q. Ayub, et al., Nucleic Acids Res. 28 (2) (2000) e8.
[5] ABI Prism 310 Genetic Analyzer. User's Manual, Applera, 2001.
[6] M. Nei, Genetics (1978) 583–590.
[7] M. Nei, Molecular Evolutionary Genetics. Columbia University Press, New York.
[8] S. Scheneider, D. Roessli, Excoffier. Arlequin ver. 2000. University of Geneve, Switzerland.
[9] J. Felsenstein, PHYLIP (version 3.2), Cladistics 5 (1989) 164–166.
[10] R.D.M. Page, Treeview, Comput. Appl. Biosci. 12 (1996) 357–358.
[11] L. Gusmão, et al., Forensic Sci. Int. 135 (2003) 150–157.
[12] L. Quintans, et al., Forensic Sci. Int. 131 (2003) 220–224.