

Analysis of 16 Y-chromosomal STRs in a Cartagena (Colombia) population sample

J.J. Builes^{a,b,*}, M.L.J. Bravo^a, A. Gómez^b, L. Caraballo^c,
C. Espinal^a, D. Aguirre^a, A. Montoya^b, M.A. Moreno^{a,b},
M.M. de Pancorbo^d, L. Gusmão^e, B. Martínez^c

^a GENES Ltda., Laboratorio de Genética Forense y Huellas Digitales del DNA, Medellín, Colombia

^b Instituto de Biología, Universidad de Antioquia, Medellín, Colombia

^c Instituto de Investigaciones Inmunológicas, Universidad de Cartagena, Cartagena, Colombia

^d Servicio de Genómica: Banco de ADN, Universidad del País Vasco, Vitoria-Gasteiz, España
^e IPATIMUP, R. Dr. Roberto Frias, s/n. 4200-465 Porto, Portugal

Abstract. We studied and established a data base of 16 Y-STR (DYS19, DYS385, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS460, DYS461, GATA-A10, GATA-H4 and DYS635) in a population of 173 unrelated males of Cartagena (Colombia) and some parameters of forensic importance were calculated. The haplotype diversity was 1.000 ± 0.0006 . This approach represents a very powerful tool for individual identification and paternity testing in forensic medicine. © 2005 Published by Elsevier B.V.

Keywords: Cartagena; Y-chromosome; STR; Population; Colombia

1. Introduction

The Y-chromosome non-recombinant portion represents a paternally inherited haploid transmission pattern [1]. Because of that Y-STRs can be employed to construct highly discriminative Y haplotypes. They are useful in stain analysis [2], paternity testing (lineage cases with male offspring) [1–3] and forensic genetics because of their male-specificity [4] and in the population genetic studies.

Here we report gene frequencies, gene and haplotype diversity for 16 Y-STR loci in the Cartagena (Colombia) population. These will increase the database and the knowledge of polymorphisms on Colombian populations.

* Corresponding author. GENES Ltda., Laboratorio de Genética Forense y Huellas Digitales del DNA, Carrera 48 No. 10-45 Cons. 611, Medellín, Colombia. Tel.: +57 4 268 48 75; fax: +57 4 318 52 70.

2. Materials and methods

Samples of 173 unrelated males were obtained from healthy individuals from Cartagena (Colombia). Genomic DNA was extracted by salting-out [5]. DNA amplification and detection of the amplicons were performed according to Builes et al. [6,7]. Alleles were identified based on the number of variable repeats and their attribution was made by comparison with an in-house constructed allelic ladder following the published nomenclature and ISFG recommendations on Y chromosome STR analysis [8]. The AMOVA, gene frequencies and gene/haplotype diversity values were calculated using the software ARLEQUIN version 2000 [9] and Nei formulation [10].

3. Results and discussion

Allele frequencies of the systems and gene diversity values are shown in Table 1. The highest diversity value in this study was found at the locus DYS385 (0.891), followed by the DYS635 (0.688). The haplotype diversity has the same value as the power of discrimination (PD) [11] and chance of exclusion (CE) [12]. The 16 STRs described in this study result in informative Y-haplotypes with CE and PD values of 1.0000.

By combining the allelic states of the 16 Y-chromosomal STR we could construct highly informative haplotypes that allowed the discrimination of 100% (173 out of 173) of the samples tested. The AMOVA results show that the percentage of variation is mainly within populations (99.95%) in agreement with previous results in European populations [13].

Development of Y-chromosome specific polymorphisms will be of great benefit in analyzing mixed DNA samples, in investigating sexual assaults as well as in paternity testing where the alleged father is not available but other patrilineal relatives are.

References

- [1] M.A. Jobling, A. Pandya, C. Tyler-Smith, *Int. J. Leg. Med.* 110 (1997) 118–124.
- [2] J. Henke, et al., *CMJ* 42 (3) (2001) 292–297.
- [3] C. Gehrig, M. Hochmeister, B. Budowle, *J. Forensic Sci.* 45 (2) (2000) 436–439.
- [4] E. Bosch, et al., *Forensic Sci. Int.* 125 (2002) 42–51.
- [5] S.A. Miller, D.D. Dykes, H.F. Polesky, *Nucleic Acids Res.* 16 (1988) 1215.
- [6] J.J. Builes, et al., *Progress in Forensic Genetics* 10 (2004) 310–312.
- [7] J.J. Builes, et al., *Progress in Forensic Genetics* 10 (2004) 275–277.
- [8] P. Gill, et al., *Forensic Sci. Int.* 124 (2001) 5–10.
- [9] S. Schneider D. Roessli L. Excoffier, University of Geneva (2000).
- [10] M. Nei, *Molecular Evolutionary Genetics*, Columbia University Press, New York, 1987.
- [11] G.F. Sensabaugh, Prentice-Hall. Englewood Cliffs (1982).
- [12] A. Chakravarti, C.C. Li, American Association of Blood Banks, Arlington, VA (1983).
- [13] L. Roewer, et al., *Forensic Sci. Int.* 114 (2000) 31–43.