International Congress Series 1288 (2006) 180–182

www.ics-elsevier.com

# AmpFℓSTR®Y-filer™: A new tool for rapid Y-STR forensic haplotyping

M.L. Pontes [a,*], D. Abrantes [a], G. Lima [a], L. Cainé [a], M.J. Pereira [a], P. Matos [a], M.F. Pinheiro [a,b]

[a] *Delegação do Porto do Instituto Nacional de Medicina Legal, Jardim Carrilho Videira, 4050-167 Porto, Portugal*

[b] *Faculdade de Ciências da Saúde-Universidade Fernando Pessoa, Portugal*

**Abstract.** Allele frequencies and population data for 17 Y-STR loci, including all the markers in the actually used European "extended haplotype" (DYS19, DYS189I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385I/II, DYS438 and DYS439) and also DYS437, DYS448, DYS456, DYS458, DYS635 and Y GATA H4 included in a new commercial kit that has recently been available, the AmpFℓSTR® Y-filer™ PCR amplification kit (Applied Biosystems), were obtained from a sample of 175 healthy unrelated males and 45 father–son pairs from the North of Portugal. A total of 171 haplotypes were identified, of which 167 were unique and 4 found in two individuals. The haplotype diversity (99. 97%) and discrimination capacity (95.43%) were calculated. © 2005 Published by Elsevier B.V.

*Keywords:* Y-chromosome; Short tandem repeat typing; Multiplex amplification; North of Portugal; Y-filer™

## 1. Introduction

In addition to the standard panel of autosomal loci used in forensic genetics, Y-STR haplotyping gives the ability to sensitive typing of male-specific DNA especially in sexual assault cases or other situations where mixtures of male and female cells are present [1]. Within the last years, a number of Y-STR multiplex assays have been developed [2]. Recently, a new commercial kit has been available, the AmpFℓSTR® Y-filer™ PCR amplification kit (Applied Biosystems) that permits the simultaneous amplification of 17 Y-STR loci, including all the markers in the actually used European "extended haplotype",

* Corresponding author. Tel.: +351 22 2073850; fax: +351 22 3325931.
*E-mail address:* mariopontes@netcabo.pt (M.L. Pontes).

0531-5131/ © 2005 Published by Elsevier B.V.
doi:10.1016/j.ics.2005.09.132

Table 1
Allele frequencies at 17 Y-STRs in a North of Portugal population sample (175 individuals)

| Allele | DYS19 | DYS389I | DYS389II | DYS390 | DYS391 | DYS392 | DYS393 | DYS437 | DYS438 | DYS439 | DYS448 | DYS456 | DYS458 | DYS635 | GATA H4 | Genotypes | DYS385 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 8 | | | | | | | | | 0.006 | 0.040 | | | | 0.029 | | 10–14 | 0.006 |
| 9 | | | | | 0.051 | | | | 0.114 | 0.337 | | | | 0.206 | 0.011 | 11–11 | 0.017 |
| 10 | | | | | 0.503 | 0.006 | | | 0.291 | 0.469 | | | | 0.086 | 0.017 | 11–12 | 0.023 |
| 11 | | 0.006 | | | 0.423 | 0.360 | | | 0.051 | 0.143 | | | | 0.520 | 0.320 | 11–13 | 0.040 |
| 12 | 0.006 | 0.166 | | | 0.023 | 0.057 | 0.206 | | 0.509 | 0.006 | | | 0.006 | 0.120 | 0.583 | 11–14 | 0.303 |
| 13 | 0.120 | 0.657 | | | | 0.537 | 0.651 | | 0.029 | | | 0.011 | 0.000 | 0.040 | 0.069 | 11–15 | 0.114 |
| 14 | 0.566 | 0.166 | | | | 0.040 | 0.120 | 0.366 | | | | 0.097 | 0.046 | | | 12–12 | 0.029 |
| 15 | 0.229 | 0.006 | | | | | 0.023 | 0.514 | | | | 0.457 | 0.097 | | | 12–13 | 0.006 |
| 16 | 0.063 | | | | | | | 0.114 | | | | 0.314 | 0.200 | | | 12–14 | 0.034 |
| 17 | 0.017 | | | | | | | 0.006 | | | 0.006 | 0.097 | 0.303 | | | 12–15 | 0.034 |
| 18 | | | | | | | | | | | 0.086 | 0.023 | 0.211 | | | 12–17 | 0.006 |
| 19 | | | | | | | | | | | 0.486 | | 0.114 | | | 12–20 | 0.006 |
| 20 | | | | | | | | | | | 0.246 | | 0.023 | | | 13–14 | 0.051 |
| 21 | | | | 0.029 | | | | | | | 0.137 | | | | | 13–15 | 0.046 |
| 22 | | | | 0.103 | | | | | | | 0.029 | | | | | 13–16 | 0.034 |
| 23 | | | | 0.194 | | | | | | | 0.011 | | | | | 13–17 | 0.040 |
| 24 | | | | 0.554 | | | | | | | | | | | | 13–18 | 0.006 |
| 25 | | | | 0.091 | | | | | | | | | | | | 13–19 | 0.011 |
| 26 | | | 0.023 | 0.017 | | | | | | | | | | | | 14–14 | 0.011 |
| 27 | | | 0.029 | 0.006 | | | | | | | | | | | | 14–15 | 0.029 |
| 28 | | | 0.131 | | | | | | | | | | | | | 14–16 | 0.023 |
| 29 | | | 0.480 | | | | | | | | | | | | | 14–17 | 0.011 |
| 30 | | | 0.246 | | | | | | | | | | | | | 14–18 | 0.006 |
| 31 | | | 0.080 | | | | | | | | | | | | | 14–19 | 0.006 |
| 32 | | | 0.029 | | | | | | | | | | | | | 15–15 | 0.017 |
| R | | | | | | | | | | 0.006 | | | | | | 15–16 | 0.006 |
| | | | | | | | | | | | | | | | | 15–17 | 0.006 |
| | | | | | | | | | | | | | | | | 15–18 | 0.011 |
| | | | | | | | | | | | | | | | | 16–16 | 0.006 |
| | | | | | | | | | | | | | | | | 16–17 | 0.011 |
| | | | | | | | | | | | | | | | | 16–18 | 0.023 |
| | | | | | | | | | | | | | | | | 16–19 | 0.011 |
| | | | | | | | | | | | | | | | | 16–20 | 0.006 |
| | | | | | | | | | | | | | | | | 17–17 | 0.006 |

R corresponds to a duplicated allele—10–12.

DYS19, DYS189I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385I/II, DYS438, DYS439 and also DYS437, DYS448, DYS456, DYS458, DYS635 and Y GATA H4. Here we report population data for a sample of 175 males and 45 father–son pairs from the North of Portugal.

## 2. Materials and methods

Blood and/or oral swabs from 175 apparently healthy and unrelated males and 45 father–son pairs from the North of Portugal were collected. DNA was extracted using the Chelex or a standard phenol–chloroform method. The loci DYS19, DYS389I and II, DYS385, DYS390, DYS391, DYS392, DYS393, DYS456, DYS458, DYS437, DYS438, DYS439, DYS448, DYS635 and Y GATA H4 were co-amplified by using the AmpFℓSTR® Y-filer™ PCR amplification Kit (Applied Biosystems) following the conditions recommended by the manufacturer (Technical Manual) and carried out using a thermocycler GeneAmp® PCR System 9700 (Applied Biosystems). The ABI PRISM® 3100 Genetic Analyzer (Applied Biosystems) was used for genetic typing. Fragment sizes were determined automatically using the Genescan® Analysis Software v. 3.7 and allele designations were based on comparison with allelic ladders included in the Y-filer™ kit. We also used some known control samples for comparison. Allele frequencies were estimated by gene counting. Gene and haplotype diversities were calculated according to Nei [3] using the Arlequin Software, version 2000 [4] and the discrimination capacity was also determined.

### 3. Results and discussion

Allele frequencies for the 17 Y-chromosome STRs are shown in Table 1. A total of 171 different haplotypes were identified, 167 of them were unique and 4 were found in two individuals. The observed haplotype diversity was $0.9997 \pm 0.0007$. In the system DYS439, we have found a duplicated allele (10–12) that needs to be sequenced. The generated data shows that the haplotype constructed using the present set of 17 Y-STR markers studied with the AmpFℓSTR® Y-filer™ kit is highly polymorphic and discriminative in the North of Portugal population. The genotyping with this kit resulted in a greater discrimination capacity when compared with the PowerPlex® Y System kit (Promega Corporation) (data not shown). This is important when regarding forensic casework analysis since it permits the genotyping of a greater set of markers using less DNA sample. No comparisons were made with other populations due to the lack of information for these particular set of loci.

In 2 of the 45 father–son pairs analysed, we have found inconsistencies of allelic transmission in the loci DYS448 and Y DYS456, which must be confirmed by sequencing.

## References

[1] M. Prinz, et al., Validation and casework application of a Y chromosome specific STR multiplex, Forensic Sci. Int. 120 (2001) 177–188.
[2] J.M. Butler, A novel multiplex for simultaneous amplification of 20 Y chromosome STR markers, Forensic Sci. Int. 129 (2002) 10–24.
[3] M. Nei, Molecular Evolutionary Genetics, Columbia Univ. Press, New York, 1973.
[4] S. Schneider, D. Roessli, L. Excoffier, Arlequin Version 2.000: A Software for Population Genetics Data Analysis, Genetics and Biometry Laboratory, University of Geneva, Switzerland, 2000.