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Allele frequencies of 15 STR loci in Argentine population

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Abstract. Short tandem repeat (STR) loci are the most informative PCR based genetic markers available to date for attempting to individualize biological material. The full use of DNA typing technology in forensic science has grown up by the development of National DNA databases. The aim of this study was to determine allele frequencies and statistical parameters of medico-legal interest, such as heterozygosity value, discrimination power and chances of exclusion in paternity cases for the loci D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D13S317, D7S820, D16S539, THO1, TPOX and CSF1PO, D2S1338 and D19S433.

Keywords: STRs; PCR; Population genetics; ABI 310; Forensics

1. Introduction

Short tandem repeat (STR) systems are highly polymorphic loci consisting of tandemly repeated sequences 1–6 bp in length. Because of the relatively small size of STR alleles (generally 100–350 nucleotides), amplification by polymerase chain reaction (PCR) is relatively easy, with the additional advantage of making the typing of forensic material successful. In addition, STR loci can be amplified simultaneously in a multiplex PCR. Thus, substantial information can be obtained in a single analysis with the benefits of using less DNA template, reducing labor and contamination. For this reason, short tandem repeat (STR) polymorphism has become a powerful tool for human identification, chromosome mapping and linkage analysis, and the study of molecular evolution and population genetics. These multiplex systems are useful for forensic identification and/or study of biological evidences and parentage testing. The STR loci are ideal candidates for co-amplification and actually we can investigate 16 loci simultaneously, 15 tetranucleotyde loci (13 CODIS loci and the loci D2S1338 Ref. [7] and D19S433 Ref. [8]) and a locus for sex identification: Amelogenin. The fluorescent multicolor dye technology allows these

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Table 1
Discrimination power and exclusion power for 15 loci

Locus	PD	PE
D8S1179	0.929	0.634
D21S11	0.941	0.647
D7S820	0.934	0.6
CSF1PO	0.866	0.425
D3S1358	0.922	0.479
TH01	0.905	0.634
D13S317	0.945	0.634
D16S539	0.914	0.647
D2S1338	0.970	0.699
D19S433	0.930	0.536
VWA	0.932	0.490
TPOX	0.775	0.250
D18S51	0.96	0.7
D5S818	0.9	0.48
FGA	0.963	0.713

multiplex loci to be analyzed on an ABI Prism® 310 or 3 100 Genetic Analyzer™ in a single capillary injection. The use of these genetic markers in forensic identification and paternity testing requires the existence of appropriate databases for the populations where the systems are going to be used. To interpret the match significance between genetically typed samples, it is necessary to know the population distribution of alleles at each locus in question.

The aim of this study was to determine allele frequencies and statistical parameters of medico-legal interest, such as heterozygosity value, discrimination power and chances of exclusion in paternity cases for the loci D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D13S317, D7S820, D16S539, THO1, TPOX and CSF1PO, D2S1338 and D19S433 (Table 1).

2. Materials and methods

Population studies were carried out in 502 unrelated individuals that came from 375 parentage tests. Genomic DNA was isolated from blood samples with salting out Miller's method [9]. The samples were amplified at the loci FGA, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D18S51 and D21S11 using the AmpFlSTR® Profiler Plus Kit (PE Biosystems, Foster City, CA) and at the loci CF1PO, TPOX, TH01, D3S1358, D7S820, and D16S539 using the AmpFlSTR® Cofiler™ kit (PE Biosystems). The same loci and the D2S1338 and D19S433 were amplified with the AmpFlSTR® Identifiler™ Kit (PE Biosystems). Samples were analyzed using the ABI PRISM® 310 Genetic Analyzer (PE Biosystems), according to the manufacturer's recommendations [1] using as separation medium Performance Optimized Polymer (POP) 4™ (PE Biosystems). The data were acquired by ABI PRISM™ 310 Collection 1.0.2 software and analyzed by GeneScan® Analysis 3.1 software and Genotyper® 2.5 according to the manufacturer's recommendations [2,4].

3. Results

All 15 loci were in Hardy–Weinberg equilibrium. Forensic statistical parameters were according to those obtained by other authors.

1. All the loci are highly polymorphics in our population. They showed heterozygosity's percentages from 57% (TPOX) to 85.9% (FGA) [3].
2. TPOX (PD = 0.77) is the less discriminator locus while D2S1338 (PD = 0.97), FGA (PD = 0.963) and D18S51 (PD = 0.96) are greater power discrimination system [3].
3. The Combined Discrimination Power is greater than 0.999999998 [3,5,6].
4. The Match Probability of all loci is 4.58466×10^{-18} [3].
5. The loci with the greatest power of exclusion are FGA and D2S1338 while TPOX has least [7].
6. The Combined Paternity Index is 922295.21 [3].
7. The Combined Exclusion Power is 0.999999 [3].
8. We have absolute concordance in the results obtained for the sample typification with the Identifiler™ Kit and the Profiler-Cofiler™ Kit, except the determination of the 18.2 allele at FGA locus.
9. We find a new allele at D13S317: 6.2 allele, it contains 197.33 bp.

4. Conclusions

This study demonstrates that this multiplex system is a useful and effective tool for forensic identification or individualization of biological evidence and parentage testing in Argentine population.

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