

Genetic population data from Araraquara region (SP State, Brazil) using PowerPlex[®] 16 Systems

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Abstract. In the present study, allele frequency distributions for the 15 STR loci included in the PowerPlex[®] 16 Systems (Promega) were obtained from a sample of 55 unrelated individuals living in Araraquara region (SP, Brazil). The frequency of each allele for each locus tested, the exact test and the forensic and paternity parameters were calculated using POWERSTATS ver. 1.2 (Promega) and GENEPOP ver. 3.2 software. All loci are in the Hardy–Weinberg equilibrium and they reached a combined power discrimination of 0.99999999999999973 and combined power exclusion of 0.99999987, showing to be a powerful tool for paternity testing and individual identification in the population analyzed. © 2005 Elsevier B.V. All rights reserved.

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1. Introduction

Because of the fast evolution in molecular biology techniques, DNA analysis is today the most sensible and specific method for human identification being extremely used in solving most of the different forensic cases and paternity tests. However, several studies demonstrate that allele frequency variations of these markers exist between different ethnic and population groups [1]. As the genetic population data of polymorphic markers are still

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Table 1
Allele frequencies and statistical parameters in a population sample of Araraquara region (SP, Brazil)

| Allele | D3S1358 | TH01 | D21S11 | D18S51 | PENTA E | D5S818 | D13S317 | D7S820 | D16S539 | CSF1PO | PENTA D | VWA | D8S1179 | TPOX | FGA |
|-------------------|---------|--------|--------|--------|---------|--------|---------|--------|---------|--------|---------|--------|---------|--------|--------|
| 2.2 | | | | | | | | | | | 0.011 | | | | |
| 5 | | | | 0.06 | | | | | | | | | | | |
| 6 | 0.202 | | | | | | | | | | | | | 0.020 | |
| 7 | 0.192 | | | 0.142 | 0.009 | | 0.045 | | 0.029 | | | | | | 0.030 |
| 8 | 0.183 | | | 0.036 | 0.018 | 0.136 | 0.191 | 0.029 | | 0.033 | | | | | 0.530 |
| 9 | 0.163 | | | 0.036 | 0.036 | 0.082 | 0.136 | 0.125 | 0.029 | 0.272 | | | | | 0.080 |
| 9.3 | 0.250 | | | | | | | | | | | | | | |
| 10 | 0.010 | | 0.009 | 0.095 | 0.055 | 0.045 | 0.218 | 0.125 | 0.221 | 0.130 | | | 0.109 | 0.060 | |
| 11 | | | | 0.095 | 0.427 | 0.300 | 0.245 | 0.317 | 0.317 | 0.130 | 0.009 | 0.091 | 0.220 | | |
| 12 | | | | 0.109 | 0.190 | 0.264 | 0.255 | 0.145 | 0.279 | 0.327 | 0.109 | | 0.136 | 0.230 | |
| 13 | | | | 0.145 | 0.107 | 0.173 | 0.136 | 0.018 | 0.096 | 0.077 | 0.196 | | 0.309 | | |
| 14 | 0.082 | | | 0.145 | 0.024 | 0.018 | 0.045 | | 0.029 | | 0.076 | 0.109 | 0.245 | | |
| 15 | 0.227 | | | 0.155 | 0.071 | | | | | | 0.043 | 0.082 | 0.100 | | |
| 16 | 0.282 | | | 0.155 | 0.036 | | | | | | | 0.218 | 0.009 | | |
| 17 | 0.255 | | | 0.145 | 0.060 | | | | | | | 0.227 | | | |
| 18 | 0.145 | | | 0.018 | 0.024 | | | | | | | 0.227 | | | 0.009 |
| 19 | | | | 0.082 | | | | | | | | 0.109 | | | 0.047 |
| 20 | 0.009 | | | 0.027 | | | | | | | | 0.018 | | | 0.132 |
| 20.2 | | | | | | | | | | | | | | | 0.019 |
| 21 | | | | 0.024 | | | | | | | | | | | 0.236 |
| 21.2 | | | | | | | | | | | | | | | 0.009 |
| 22 | | | | | | | | | | | | | | | 0.151 |
| 23 | | | | | | | | | | | | | | | 0.170 |
| 24 | | | | | | | | | | | | | | | 0.104 |
| 24.2 | | 0.009 | | | | | | | | | | | | | |
| 25 | | | | | | | | | | | | | | | 0.075 |
| 26 | | 0.009 | 0.009 | | | | | | | | | | | | 0.038 |
| 27 | | 0.036 | | | | | | | | | | | | | 0.009 |
| 28 | | 0.245 | | | | | | | | | | | | | |
| 29 | | 0.145 | | | | | | | | | | | | | |
| 30 | | 0.264 | | | | | | | | | | | | | |
| 30.2 | | 0.018 | | | | | | | | | | | | | |
| 31 | | 0.064 | | | | | | | | | | | | | |
| 31.2 | | 0.118 | | | | | | | | | | | | | |
| 32 | | | | | | | | | | | | | | | |
| 32.2 | | 0.064 | | | | | | | | | | | | | |
| 33.2 | | 0.027 | | | | | | | | | | | | | |
| <i>Parameters</i> | | | | | | | | | | | | | | | |
| Ho | 0.7455 | 0.8909 | 0.8462 | 0.8727 | 0.9048 | 0.6909 | 0.8545 | 0.8545 | 0.7885 | 0.6731 | 0.8478 | 0.8182 | 0.8545 | 0.6400 | 0.8679 |
| He | 0.7833 | 0.8320 | 0.8073 | 0.8769 | 0.9068 | 0.7196 | 0.8045 | 0.8210 | 0.7935 | 0.7431 | 0.8423 | 0.8257 | 0.8026 | 0.6624 | 0.8607 |
| Exact test | 0.4656 | 0.4417 | 0.7537 | 0.9177 | 0.3039 | 0.2853 | 0.4143 | 0.1969 | 0.3717 | 0.7137 | 0.7786 | 0.1401 | 0.7804 | 0.8027 | 0.1986 |
| PD | 0.9091 | 0.9135 | 0.9256 | 0.9600 | 0.9592 | 0.8714 | 0.9177 | 0.9144 | 0.9149 | 0.8883 | 0.9386 | 0.9243 | 0.9157 | 0.8496 | 0.9434 |
| PE | 0.5020 | 0.6873 | 0.7769 | 0.7401 | 0.8052 | 0.4143 | 0.7038 | 0.7038 | 0.5432 | 0.3879 | 0.6906 | 0.6332 | 0.7038 | 0.3417 | 0.7685 |
| MP | 0.0909 | 0.0865 | 0.0744 | 0.0400 | 0.0408 | 0.1286 | 0.0823 | 0.0856 | 0.0851 | 0.1117 | 0.0614 | 0.0757 | 0.0843 | 0.1504 | 0.0566 |
| PIC | 0.7398 | 0.7681 | 0.8028 | 0.8544 | 0.8872 | 0.6698 | 0.7691 | 0.7868 | 0.7475 | 0.6902 | 0.8130 | 0.7931 | 0.7669 | 0.6186 | 0.8383 |
| TPI | 1.96 | 3.25 | 4.58 | 3.93 | 5.25 | 1.62 | 3.44 | 3.44 | 2.17 | 1.53 | 3.29 | 2.75 | 3.44 | 1.39 | 4.42 |

Ho: heterozygosity observed, He: heterozygosity expected, PD: power of discrimination, PE: power of exclusion, MP: matching probability, PIC: polymorphism information content, TPI: typical paternity index.

unknown in Brazilian population, the aim of this work was to study allele frequency distributions of the 15 STR loci using PowerPlex® 16 Systems (Promega) in the population of Araraquara region (SP, Brazil) and to report some statistical parameters of forensic and paternity interest.

2. Materials and methods

Blood samples were obtained from 55 unrelated individuals living in Araraquara region. DNA was extracted using GenomicPrep™ Blood DNA Isolation kit (Amersham).

The amplification was performed using PowerPlex® 16 Systems kit (Promega) in a PTC-100 PCR Systems (MJ Research), following the manufacturer's recommendations. The amplified products were run on denaturing 6% polyacrylamide gel in an ABI PRISM® 377 DNA Sequencer (Applied Biosystems) and analysed with the GeneScan ver. 2.1 analysis software (Applied Biosystems).

The frequency of each allele for each locus tested was calculated using the number of observed genotypes in the sample by POWERSTATS (Promega) software ver. 1.2 [2]. The exact test for the Hardy–Weinberg equilibrium as carried out using GENEPOP software ver. 3.2 [3] and the forensic and paternity parameters (Ho: Heterozygosity observed, He: Heterozygosity expected, PD: Power of Discrimination, PE: Power of Exclusion, MP: Matching Probability, PIC: Polymorphism Information Content, TPI: Typical Paternity Index) were performed using POWERSTATS (Promega) ver. 1.2 [2] and GENEPOP ver. 3.2 [3] software.

3. Results and discussion

The distribution of the alleles for all 15 STR loci is shown in Table 1. After all calculations, it was observed that no deviation from Hardy–Weinberg equilibrium was detected for all markers in this study. Moreover, all loci were highly polymorphic and loci as PENTA E (90.5%), TH01 (89.0%), D18S51 (87.2%) and FGA (86.8%) had the highest observed heterozygosities, while the locus TPOX (64%) showed the lowest observed heterozygosity. TPOX also presented lower discrimination power (85.0%) and D18S51 (96.0%), PENTA E (95.92%) and FGA (94.3%) were greater discrimination power systems. The combined power exclusion was 0.99999987, ranging from 0.34 (TPOX) to 0.81 (PENTA E) and the combined power discrimination was 0.99999999999999973, ranging from 0.85 (TPOX) to 0.96 (D18S51).

4. Conclusion

These results indicated that the 15 loci studied would be useful as genetic markers for forensic identification and paternity testing in Araraquara region population (SP, Brazil).

References

- [1] L. Pastore, et al., Allele frequency distributions at several variable number of tandem repeat (VNTR) and short tandem repeat (STR) loci in a restricted Caucasian population from South Italy and their evaluation for paternity and forensic use, *Mol. Cell. Probes* 10 (1996) 299–308.
- [2] <http://www.promega.com/geneticidtools/powerstats/Default.htm>.
- [3] M. Raymond, F. Rousset, GENEPOP (version 1.2): populations genetics software for exact tests and ecumenicism, *J. Heredity* 86 (1995) 248–249.