



Allele frequencies of three forensic STR markers (D22S1045, D2S441 and D10S1248) in the population from São Paulo, Brazil



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ABSTRACT

São Paulo state is one of the Brazilian states with the highest cultural and ethnic miscegenation, mainly of Italian and Portuguese descendants, but also strongly influenced by Amerindians, Africans, Spanish, Germans, Arabs and Japanese, as well as the high number of immigrants from the northeast region. In this work we analyzed statistical data of three STRs markers (D22S1045, D2S441 and D10S1248) that are used in our laboratory's forensic routine for paternity investigations. The use of these data will be extremely important for the laboratory to perform statistical analyzes for the conclusion of the reports. We analyzed 221 samples obtained from unrelated individuals born in the São Paulo state, which the allele frequencies and statistical parameters were estimated with PowerStats version 12 (Promega Corp.). The power of discrimination (PD) and power of exclusion (PE) for the D10S1248 marker were 0.914 and 0.576 respectively and the allele 14 showed the highest frequency (0.294) and 8, 10, 18 and 19 alleles presented the lowest frequency (0.002). PD and PE of the D2S441 marker were 0.907 and 0.551 respectively; the highest frequency allele was 11 (0.312) and the lowest frequency alleles were 9 and 12.3 (0.002). The marker D22S1045 showed PD 0.890 and PE 0.489 with a higher frequency allele 16 (0.373) and lowest frequency allele 8 (0.002). Also, significant differences were found between the São Paulo population and the other populations for the three markers analyzed.

1. Introduction

Short Tandem Repeat (STR) polymorphisms are widely used in forensic analysis. Common cases of paternity are analyzed by commercially available multiplex systems, however, for more complex cases, such as complex kinship analysis, additional STRs are required for best results [1].

In order to increase the amount of frequency data and genetic parameters of the Brazilian population, three STRs (D22S1045, D2S441 and D10S1248) were analyzed and used in the forensic routine of our laboratory for paternity investigation. The majority individuals of these cases come from the São Paulo' state, Brazil, which is considered the Brazilian state with the most cultural and ethnic miscegenation, mainly comprising of Italians and Portuguese descendants, but also with strong influence of Amerindians, Africans, Spanish, Germans, Arabs and Japanese populations, as well as a high number of immigrants from the northeast region [2]. Considering this particularity, the use of regional frequency data will be extremely important for the laboratory, facilitating the conclusion of the reports.

2. Materials and methods

A total of 221 samples obtained from unrelated individuals born in the São Paulo state were collected on FTA® classic cards (GE Healthcare, Life Sciences). Samples were amplified with the PowerPlex® Fusion 6C System (Promega), following the User Guide [3]. Capillary electrophoresis and detection were performed on the 3500 Genetic Analyzer using POP-4® polymer (Applied Biosystems). Genotypes were assigned using the GeneMapper ID v1.2.1 software (Applied Biosystems). Allele frequencies and statistical parameters were estimated with PowerStats version 12 (Promega Corp.) [4]. Allelic frequencies for each locus in São Paulo (Southeast Brazil) population were compared with other populations: European, African [5], Colombian region [6] and the population of the Rio Grande do Sul' state (Southern Brazil) [7]. Pairwise genetic distances were calculated between populations using Arlequin v3.5 [8] software. Our laboratory successfully participated in proficiency testing provided by the GEP-ISFG Working Group.

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Table 1
Allele frequencies and forensic parameters of the three STR loci for São Paulo population, Southeast Brazil.

Allele	D22S1045	D2S441	D10S1248
8	0,0023		0,0023
9		0,0023	
10	0,0113	0,1742	0,0023
11	0,1176	0,3122	0,0113
11.3		0,0566	
12	0,0158	0,0814	0,0588
12.3		0,0023	
13	0,0068	0,0362	0,2579
14	0,0362	0,3009	0,2941
15	0,2986	0,0294	0,1968
16	0,3733	0,0045	0,1471
17	0,1267		0,0249
18	0,0113		0,0023
19			0,0023
N	442	442	442
HO	0,262	0,226	0,213
HE	0,738	0,774	0,787
PIC	0,700	0,735	0,749
PD	0,890	0,907	0,914
PE	0,489	0,551	0,576

N: number of chromosomes; Ho: observed heterozygosity; He: expected heterozygosity; PIC: polymorphism information content; PD: power of discrimination; PE: power of exclusion.

Table 2
Comparison of the allele frequencies for D2S441, D10S1248 and D22S1045 loci between the São Paulo State population and other populations.

Microsatellite markers	Southern Brazil × São Paulo	Europe × São Paulo	África × São Paulo	Colombian × São Paulo
D2S441	0.19992	0.17631	0.17925	–
D10S1248	0.00982	0.13288	0.10796	0.12997
D22S1045	0.13122	0.12334	0.10917	0.15853

– no data available.

3. Results and discussion

The power of discrimination (PD) and power of exclusion (PE) for the marker D10S1248 were 0.914 and 0.576 respectively; the highest frequency allele was 14 (0.294) and the lowest frequency were 8, 10, 18 and 19 (0.002) (Table 1). PD and PE of the marker D2S441 were 0.907 and 0.551, respectively; the allele with the highest frequency was 11 (0.312) and the lowest frequency were 9 and 12.3 (0.002). The D22S1045 marker showed PD 0.890 and PE 0.489, highest frequency in 16 allele (0.373) and lowest allele 8 (0.002).

The analysis of genetic distance showed significant differences between São Paulo population from Europe, Africa, Colombia and Southern Brazilian region. The marker that presented the greatest genetic distance among the populations was D2S441 (Table 2).

4. Conclusion

These 3 STRs loci increase the discriminatory power of forensic analysis and are very useful for forensic purposes. Among the analyzed, the marker with the greatest power of discrimination for the population of São Paulo was D10S1248.

Conflict of interest statement

None.

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