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Power of exclusion of 18 autossomic STR loci in a Brazilian Middle–West region population sample

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Abstract. The aim of this work was to analyze the Power of Exclusion (PE) in paternity investigation cases from Federal District of Brazil (Center–West region of Brazil) using a local population sample ($n=917$) in comparison with Promega Corporation published databank, both for 18 autossomic STR loci. For this purpose, we analyzed 311 cases where the alleged father was excluded. The expected and observed values in the exclusion cases were compared by *locus* and by multiplex system. The values of χ^2 show a P higher than 0.05 for each *locus*, except TPOX and D16S539 ($0.05>P>0.01$), where the observed number of exclusions was higher than the expected ones. The a priori Exclusion Probability to each *locus* did not differ significantly between the two databases, suggesting that, in the absence of a database population-specific, another databank can be used as reference for estimates of PE and Probability of Paternity, considering similar ancestry in both populations composition, which is the case of Brazil and the United States.

Keywords: Power of exclusion; Autossomic short tandem repeat locus; Brazilian population

1. Introduction

In a classical analysis of genetic relationship, one of the useful parameters is the Power of Exclusion (PE)—the power of a genetic marker in excluding a non-related individual, chosen by chance in a specific population, as an alleged father in a paternity investigation. The paternity PE is the expected average probability that a polymorphic *locus* shows the exclusion of a man without kinship with the biological father. This index depends on the informative content of a *locus*, which depends on its number of alleles and its respective

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frequencies. From the probabilities of exclusion of several *loci*, it is possible to calculate the Combined PE (PEC), by simple multiplication of the values for each *locus*. The value of PEC is function of the examined *loci* number, as well as of the informative content of each *locus*. The knowledge of the PE and the PEC can define the *loci* to be used in an analysis of genetic relationship [1]. Microsatellite *loci* are usually adopted by its high polymorphism, which allows the individualization of a person when combined.

The purposes of this work were: (1) analyze the PE of 18 STR (*short tandem repeat*) *loci*, included in three commercial multiplex systems–PowerPlex® 1.1 (PP1.1) [2], PowerPlex® 2.1 (PP2.1) [3] and FFFL [4]–employed by the Instituto de Pesquisa de DNA Forense (IPDNA) of the Judiciary Police of the Federal District of Brazil in paternity cases; (2) analyze the PE of these multiplex systems employing two databanks: IPDNA [5] and North-American sample databank (Promega Corporation).

2. Material and methods

The IPDNA databank consists of 917 non-related individuals, representing a random sample of the Federal District (Middle–West region of Brazil, where the country capital is located). The analyzed samples were obtained from paternity investigation cases (only the parents' genotypes were considered) and criminal investigation from the Federal District Justice. Three multiplex amplification systems were used: PP 1.1, PP 2.1 and FFFL (in a total of 18 STR *loci*: Penta E, D18S51, D21S11, D3S1358, FGA, vWA, D16S539, D7S820, D5S818, D16S539, D7S820, D13S317, D5S818, CSF1PO, F13A01, FESFPS, F13B e LPL) [5].

From 1188 paternity investigation cases with established trio evaluated, 311 resulted in the alleged father exclusion, each with at least 3 non-inclusion events. The times that each *locus* contributed in the exclusion characterization (observed exclusions) was compared with the expected ones based on the PE of each *locus* (established from the databases). In consequence of the interval range of the sample collections, the number of cases where each genetic marker was analyzed showed variation.

In the paternity inquiries, the IPDNA searchs a PE (as well as the Probability of Paternity) of at least 99.99%. For such a way, the minimum of regions needed was analyzed to reach such values, according to the multiplex systems (PP1.1, PP2.1 and FFFL), separately, two by two and all three together. The a priori PE was obtained using the formula: $PE = h^2[1 - 2h(1-h)^2]$ [6], where h is the observed frequency of heterozygotes in the Federal District databank sample ($n = 917$) [5]. The PE gotten for each *locus* was compared with the values of the Promega Corporation [2–4]. The expected exclusions, estimated using the PE for each *locus* and for each multiplex system, had been compared with the observed ones in all exclusion cases, using the chi-square test from both population samples.

3. Results and discussion

According to the results (full data available under request to the corresponding author), the a priori Probability of Exclusion for each *locus* did not differ significantly between the two databanks [2–5]. It seems viable, therefore, when there is no specific databank for the studied population, the use of another one whose composition is similar in terms of ethnic origin, without significant variation in the results.

Table 1
Power of exclusion of STR markers obtained in this study and by Promega Corporation

System	Power of exclusion	
	Present study	Promega Corporation
FFFL[a]	0.92170492	0.92966667
PP 1.1[b]	0.99783306	0.99760342
PP 2.1[c]	0.99992948	0.99990755
PP 1.1+FFFL	0.99983034	0.99983144
PP 2.1+FFFL	0.99999448	0.99999350
PP 1.1+PP 2.1	0.99999831	0.99999800
PP 1.1+PP 2.1+FFFL	0.99999987	0.99999986

[a] F13A01, FESFPS, F13B, LPL.
[b] D16S539, D7S820, D13S317, D5S818, CSF1PO, TPOX, TH01, vWA.
[c] Penta-E, D18S51, D21S11, TH01, D3S1358, FGA, TPOX, D8S1179, vWA.

The analysis of the paternity exclusion cases showed that the observed exclusions for each *locus* was similar to the expected ones, considering a significance level of 5%, except in TPOX and D16S539 (P values inside the expected for a significance level of 1%), where the number of observed exclusions was greater than the expected.

Regarding the multiplex systems PE, the values of both databanks were similar, coming closer as the combination of regions increases (Table 1). The commercial system PP2.1, that includes 9 STR regions, reaches a PE of 99.999%, sufficient for the conclusion of paternity cases. This showed that PP2.1 multiplex system [3], that is a nine *loci* set, is efficient and enough for the analysis of paternity cases, making possible, in its majority, the attainment of a PE superior to 99.99%, consequence of the *loci* combination, with high resolution power and polymorphic index.

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