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Analysis of Penta D and Penta E STR loci in a Northern Portuguese population

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Abstract

Allele and genotype frequency data for two STR loci, Penta D and Penta E, were obtained from a sample of 291 unrelated inhabitants from Northern Portugal, with a commercial PCR-based typing kit. No significant deviations from Hardy–Weinberg equilibrium were found. There was no evidence for correlation between alleles of the two loci. Appropriate statistical evaluations revealed that these systems have a high forensic efficiency and can be very useful for personal identification among the Northern Portuguese population.
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1. Introduction

The profiling of highly polymorphic short tandem repeat (STR) loci has been, during the last decade, a relatively simple but powerful tool to use in population genetics studies and forensic identification [2,6,15].

However, allelic composition determination and statistical evaluations must be carried out in the populations of interest, to make later use of the studied STRs in forensic casework [2]. At the present time, there is a considerable amount of allelic data of STR markers from a great variety of populations [1,4,5,8,10,12,15].

The purpose of this study is to report allele and genotype frequency data for the Penta D and Penta E STRs in a Northern Portuguese population sample as well as statistical

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evaluations, including the possible divergence from Hardy–Weinberg expectations and other forensic parameters of interest.

2. Materials and methods

Blood samples were obtained by venipuncture of 291 unrelated individuals from Northern Portugal, using EDTA as anticoagulant.

Table 1
Allele frequencies for loci Penta D and Penta E

Penta D	
Allele	Frequency
2.2	0.0120
3	0.0017
7	0.0052
8	0.0240
9	0.1804
10	0.1220
11	0.1564
12	0.1839
13	0.1787
14	0.1082
15	0.0223
16	0.0052
Exact test $p = 0.3626$	

Penta E	
Allele	Frequency
5	0.0739
7	0.1512
8	0.0172
9	0.0051
10	0.0945
11	0.1220
12	0.1890
13	0.0997
14	0.0739
15	0.0447
16	0.0361
17	0.0395
18	0.0137
19	0.0189
20	0.0052
21	0.0103
22	0.0034
23	0.0017
Exact test $p = 0.5580$	

DNA extraction was performed by the Chelex-100 method [16].

PCR amplification was carried out using the Powerplex®16 System amplification kit (Promega) and a GeneAmp PCR System 9600 (PE Biosystems), according to the kit manufacturer's instructions [13]. The Powerplex®16 System is a commercially available kit that can be used to co-amplify 16 loci, including 13 tetranucleotide STR loci, Amelogenin and 2 pentanucleotide STR loci—Penta D and Penta E [13].

The PCR products were mixed with deionised formamide (Ultra Pure Grade, Amresco®) and Internal Lane Standard (ILS) 600, and then denatured at 95 °C for 3 min, as described in the Technical Manual [13].

All PCR product separations were performed by capillary electrophoresis using an ABI Prism 310 Genetic Analyzer (PE Biosystems). Data Collection software parameters are indicated in the Technical Manual [13].

The results were analysed with GeneScan 2.1 analysis software, and test sample allele designations were decided by comparison with the allelic ladder included in each kit.

Allele frequencies were estimated by direct counting.

Hardy–Weinberg equilibrium assessment (exact test [9]), heterozygosity calculations, as well as the pair-wise comparison of the two loci were executed with the Genepop [14] software.

Some statistical parameters of forensic interest such as the probability of exclusion (Pex [11]), the polymorphism information content (PIC [3]) and the discrimination power (PD [7]) were also calculated.

3. Results and discussion

The distributions of observed allele frequencies for the two loci Penta D and Penta E and the *p*-values of the Hardy–Weinberg exact test [9] are summarized in Table 1. The values of statistical parameters with forensic interest are shown in Table 2. According to the results obtained, three main conclusions could be drawn.

- The distribution of the genotypes of the two systems was found to be in Hardy–Weinberg equilibrium (exact test).
- There was no evidence for correlation between the alleles for the pair-wise comparison of the two loci ($p = 0.8497$).
- The statistical parameters with forensic interest revealed that these systems have a high forensic efficiency and can be very useful for personal identification in the Northern Portuguese population (Table 2).

Table 2
Statistical parameters of forensic interest

	Heterozygosity	Pex	PIC	PD
Penta D	0.8508	0.6955	0.8309	0.9588
Penta E	0.8924	0.7811	0.8812	0.9781

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