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Portuguese population data on two pentanucleotide STR loci Penta E and Penta D

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Abstract

Two pentanucleotides—Penta E and Penta D—have been studied in 160 Portuguese Caucasian individuals and in 19 African origin individuals when the GenePrint® PowerPlex™ 16 System were introduced for paternity studies in our laboratory. Allele frequencies and forensic parameters were determined in these two populations. One-hundred fifty meioses were also studied and a paternal genetic inconsistency in Penta E was detected. Furthermore, 81 selected paternity investigation cases, including 60 nonexclusion cases, 14 exclusion cases and 7 mutation cases were studied with SGM Plus and PowerPlex 16. In nonexclusion cases, paternity probability (W) values were always >99.99%, even in mutation cases including the respective mutation rate. These data point to the conclusion that Penta E and Penta D are valuable STR loci for paternity investigation in the Portuguese population.

Keywords: Penta E; Penta D; STR loci; Paternity probability; Mutation case

1. Introduction

For paternity investigation cases, we normally use SGM Plus and PowerPlex16. These two multiplex systems type a total of 17 routine STR loci including the 13 core CODIS loci, two tetranucleotide loci—D2S1338 and D19S433, and two pentanucleotide loci—Penta E and Penta D [1,2]. The two pentanucleotides were studied in 160 Portuguese Caucasian individuals and in 19 individuals of African origin from Cabo Verde. Paternity studies were performed in 81 selected paternity cases, including 60 nonexclusion cases, 14 exclusion cases and 7 mutation cases to determine the forensic value of Penta E and Penta D in these cases. Maternal and paternal meioses were also studied to detect any genetic inconsistencies.

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2. Material and methods

Penta E and Penta D have been studied in 160 unrelated Portuguese Caucasian individuals mainly from South Portugal and in 19 African origin individuals from Cabo Verde. Blood samples were collected in Ultra Stain Card, extracted with Chelex and purified with the Wizard DNA Clean-Up System. Amplifications were performed with a Perkin-Elmer GeneAmp PCR System 9600 and electrophoresis were carried out on a ABI Prism 377 DNA sequencer, according to the manufactures' instructions for SGM Plus and PowerPlex 16.

3. Results

Allele frequency distributions are shown in Table 1 and Fig. 1 for 160 Penta E and 151 Penta D samples from a Portuguese Caucasian population and for 19 Penta E and 17 Penta

Table 1
Allele frequencies for Penta D and Penta E in a Portuguese Caucasian population and in an African origin population from Cabo Verde

Population				
Alleles	Portuguese Caucasian		African origin	
	Penta D	Penta E	Penta D	Penta E
2.2			0.1176	
5	0.006	0.0844	0.0294	0.0789
6				
7	0.0033	0.1219	0.0588	0.0526
8	0.0133	0.0219	0.1176	0.2105
9	0.2133	0.0031	0.1765	0.0263
10	0.0900	0.1156	0.1471	0.0263
11	0.1733	0.1250	0.1176	0.0263
12	0.1533	0.2156	0.1177	0.1578
13	0.2266	0.0844	0.0588	0.1578
14	0.0833	0.0375	0.0588	0.0789
15	0.0300	0.0438		0.0526
16	0.0066	0.0500		0.0263
17		0.0344		0.0536
18		0.0188		
19		0.0313		
20		0.0063		
21		0.0031		0.0263
22				0.0263
23		0.0031		
E C	0.6898	0.7892	0.7576	0.7641
P D	0.9506	0.9771	0.9737	0.9749
Het	0.8362	0.8893	0.8837	0.8840

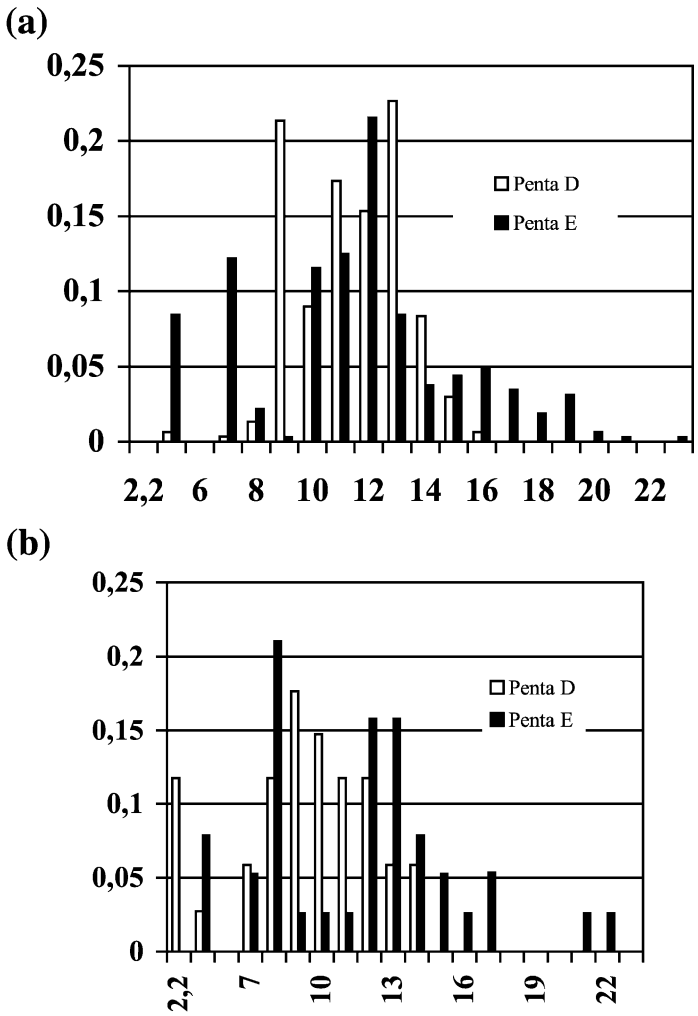

Fig. 1. Allele frequency distribution in a Portuguese Caucasian population (a) and in an African origin population from Cabo Verde (b).

D samples from a Cabo Verde African origin population. A paternal genetic inconsistency in Penta E was detected when 80 maternal and 70 paternal meioses were studied in both STR loci (Table 2).

Table 2
A paternal genetic inconsistency detected in Penta E

Loci	W without mutation	A Father	Mother	Child	W with mutation
Penta E	99.999995%	8–13	11–14	14–14	99.998%

4. Conclusions

Despite the small number of African samples studied, differences were found in allele frequencies between the African and the Portuguese populations in allele 8 and allele 22 for Penta E and in allele 2.2 for Penta D. Forensic parameters—exclusion chance (EC), power of discrimination (PD) and heterozygosity (Het)—are very high in both populations.

Considering paternity studies and examining 17 STR loci, there is a considerable difference in the number of genetic inconsistencies between exclusion cases with more than eight genetic inconsistencies (exclusions) per case and mutation cases with one or two genetic inconsistencies per case. Paternity probability values (W) were also different for mutation cases (W>99.99%) and for exclusion cases (W < 99.9%). These data point to the conclusion that Penta E and Penta D are valuable STR loci for paternity investigation in the Portuguese population.

References

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