



Gene frequencies of six miniSTR in Tuscany (Italy)

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Abstract. Six previously characterized STR loci with amplification products smaller than 125 bp have been typed in a sample of 100 unrelated subjects born in Tuscany, Italy, using a protocol involving two triplexes. These loci, called “miniSTR”, represent promising tools for recovering genetic information from degraded DNA samples when standard loci generate partial profiles.
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1. Introduction

In a recent work [1], six new STR loci generating amplicons smaller than 125 bp have been characterized in three ethnic groups from the USA. These markers (D1S1677, D2S441, D4S2364, D10S1248, D14S1434, and D22S1045) are unlinked to the CODIS loci, and were designed to be amplified in two triplexes. Here, we applied the same protocol to a sample of 100 subjects of self-reported Caucasian ancestry born in our region (Tuscany), and compared the distribution of the allele frequencies with those reported [1].

2. Materials and methods

DNA was extracted from buffy coats using the QIAamp DNA blood kit (Qiagen, Hilden, Germany); electrophoresis was carried out in the ABI 310 automated sequencer (Applied Biosystem) following standard protocols. The amplification reaction followed [1], with minor modifications.

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Table 1

Allele frequencies and expected heterozygosity of six miniSTR loci among 100 Italians

Allele	D10S1248	D14S1434	D22S1045	D1S1677	D2S441	D4S2364
8			0.130			0.010
9			0.015	0.010	0.010	0.135
10			0.010	0.010	0.170	0.560
11			0.065	0.105	0.340	0.290
11.3					0.040	
12			0.360	0.250	0.055	0.005
12.3					0.005	
13	0.025		0.355	0.370	0.030	
14	0.230	0.140	0.065	0.200	0.325	
15	0.325	0.060		0.050	0.025	
16	0.165	0.025		0.005		
17	0.215	0.350				
18	0.040	0.395				
19		0.030				
Heterozygosity	0.766	0.697	0.719	0.747	0.744	0.584
PE standard trios	0.545	0.450	0.484	0.522	0.521	0.317
PE deficiency cases	0.366	0.282	0.311	0.345	0.345	0.175

For naming the alleles, we used the control sample 9947A (Applied Biosystem) as a reference, whose reading was kindly provided by Michael Coble (NIST-Biotechnology Division). A ladder was composed by pooling selected individuals from our sample.

Statistical analyses were performed using Arlequin 2000 [2].

3. Results and discussion

One hundred unrelated individuals born in Tuscany were successfully typed. No significant deviation from Hardy-Weinberg equilibrium (permutation test) was observed at any locus. Allele frequencies, expected heterozygosities, and power of exclusion (PE) are shown in Table 1. The allele frequency distributions were similar to those observed in the US white population [1].

References

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