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Genetic study on 10 STR loci in the Romanian population

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Abstract. A population study on 10 tetrameric STR loci (D3S1358, VWA, D8S1179, D21S11, D18S51, D16S539, D2S1338, D19S433, THO1 and FGA) was performed on 104 unrelated individuals from Romania (Bucharest area), by mean of multiplex PCR (AmpFl STR SGM plus) and capillary electrophoresis. The allele frequencies and the usual forensic statistical parameters were defined. A brief comparison of the allele frequency distributions with other available data from European populations is presented. All the analyzed loci met Hardy–Weinberg equilibrium expectations. The high discrimination and exclusion power of the combined system showed the forensic efficiency of these 10 genetic markers. The study is considered to be very useful for paternal lineage testing and identification purposes in current forensic practice with Romanian individuals.

Keywords: Short tandem repeats; Romanian population; Genetic polymorphisms

1. Introduction

Polymorphic STR loci are intensively used as markers for personal identification and paternity testing. The scientific forensic genetic community is interested in compiling STRs data in different populations in order to establish the ethnic and geographic allele frequency distributions needed for the forensic casework. Extensive population studies were carried out on STRs in various European countries [1]. Only limited data on STR markers in Romanian population is available in the literature so far [2,3]. Therefore, the purpose of this study is to deliver population genetic data on 10 STR loci regarding the Romanian population. For this purpose, a population sample of 104 unrelated individuals from Romania was selected to be genotyped in a joint research project.

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Table 1
The allele frequencies, statistical parameters and the HWE analysis for 10 STR loci in the Romanian population sample (Bucharest area) ($N = 104$ individuals)

Locus alleles	TH01 ($N = 104$)	D16S359 ($N = 104$)	D8S1179 ($N = 104$)	D18S51 ($N = 104$)	D19S433 ($N = 104$)	VWA ($N = 104$)	D3S1358 ($N = 104$)	D2S1338 ($N = 104$)	FGA ($N = 104$)	D21S11 ($N = 104$)
5	0.005	–	–	–	–	–	–	–	–	–
6	0.264	–	–	–	–	–	–	–	–	–
7	0.149	–	–	–	–	–	–	–	–	–
8	0.125	0.019	0.024	–	–	–	–	–	–	–
9	0.144	0.139	0.014	–	–	–	–	–	–	–
9.3	0.308	–	–	–	–	–	–	–	–	–
10	0.005	0.077	0.053	0.005	–	–	–	–	–	–
11	–	0.284	0.087	0.010	0.014	–	–	–	–	–
12	–	0.322	0.139	0.135	0.115	–	–	–	–	–
13	–	0.120	0.356	0.159	0.197	0.005	–	–	–	–
13.2	–	–	–	–	0.019	–	–	–	–	–
14	–	0.038	0.197	0.115	0.317	0.144	0.125	–	–	–
14.2	–	–	–	–	0.029	–	–	–	–	–
15	–	–	0.087	0.154	0.120	0.106	0.245	–	–	–
15.2	–	–	–	–	0.058	–	–	–	–	–
16	–	–	0.038	0.163	0.063	0.183	0.231	0.034		–
16.2	–	–	–	–	0.053	–	–	–	–	–
17	–	–	0.005	0.125	–	0.274	0.188	0.245	–	–
17.2	–	–	–	–	0.005	–	–	–	–	–
18	–	–	–	0.053	0.005	0.216	0.192	0.101	0.019	–
18.2	–	–	–	–	0.005	–	–	–	–	–
19	–	–	–	0.019	–	0.053	0.019	0.125	0.082	–
20	–	–	–	0.019	–	0.019	–	0.135	0.091	–
21	–	–	–	0.024	–	–	–	0.034	0.192	–
22	–	–	–	0.010	–	–	–	0.014	0.192	–
22.2	–	–	–	–	–	–	–	–	0.014	–
23	–	–	–	0.010	–	–	–	0.111	0.125	–
23.2	–	–	–	–	–	–	–	–	0.029	–
24	–	–	–	–	–	–	–	0.111	0.149	–
24.2	–	–	–	–	–	–	–	–	0.005	–
25	–	–	–	–	–	–	–	0.063	0.067	–
26	–	–	–	–	–	–	–	0.029	0.019	–
27	–	–	–	–	–	–	–	–	0.014	0.029
28	–	–	–	–	–	–	–	–	–	0.106
29	–	–	–	–	–	–	–	–	–	0.221
30	–	–	–	–	–	–	–	–	–	0.188
30.2	–	–	–	–	–	–	–	–	–	0.072
31	–	–	–	–	–	–	–	–	–	0.038
31.2	–	–	–	–	–	–	–	–	–	0.106
32	–	–	–	–	–	–	–	–	–	0.024
32.2	–	–	–	–	–	–	–	–	–	0.154
33.2	–	–	–	–	–	–	–	–	–	0.063
Hobs	0.807	0.837	0.798	0.865	0.865	0.760	0.865	0.789	0.846	0.875
Hexp	0.781	0.778	0.799	0.877	0.825	0.814	0.802	0.869	0.871	0.862
MEC	0.564	0.568	0.616	0.743	0.659	0.623	0.597	0.731	0.733	0.716
MEP	0.563	0.559	0.597	0.749	0.647	0.624	0.603	0.732	0.736	0.719
PIC	0.742	0.741	0.771	0.859	0.801	0.783	0.767	0.850	0.852	0.842
PM	0.093	0.095	0.071	0.039	0.061	0.064	0.087	0.043	0.040	0.042
D	0.907	0.905	0.929	0.961	0.939	0.936	0.913	0.957	0.960	0.958
HWE test (p-values)										
Exact test	0.700	0.766	0.821	0.251	0.709	0.824	0.630	0.242	0.267	0.940

Hobs (observed heterozygosity), Hexp (expected heterozygosity), (MEC) mean exclusion chance, (MEP) mean paternity exclusion probability, (PIC) polymorphism information content, (PM) match probability, (D) discrimination power, probability values p (Hardy – Weinberg equilibrium exact test based on 5000 shuffling).

2. Material and methods

Whole EDTA blood samples was collected from 104 unrelated Romanian individuals, randomly selected among the patients of a Bucharest hospital. Appropriate informed consent was obtained from the participants. Members of the Romanian minorities (Hungarians, Germans, Gypsies, etc.) were excluded from the study.

DNA was Chelex-extracted from peripheral blood samples as previously described by Ref. [4]. Amplification was carried out using about 1 ng of template DNA in a final volume of 12.5 μl containing 1 μl of Chelex extract. All the samples have been typed for a set of 10 STR autosomal loci loci D3S1358, VWA, D8S1179, D21S11, D18S51, D16S539, D2S1338, D19S433, THO1 using an AmpFlSTR SGM plus system kit (Applied Biosystems), according to the manufacturer's recommendations. The PCR products were separated by capillary electrophoresis on ABI PRISM 310 Genetic Analyzer (Applied Biosystems) and data were analyzed using Genotyper® 2.5 software (Applied Biosystems) with the template included in the kit. The alleles were labeled according to the international nomenclature [5].

The allele frequencies were calculated from each genotype in the sample set. The statistical parameters of forensic importance (observed and expected heterozygosity, mean paternity exclusion chance, mean exclusion probability, polymorphism information content, discrimination power) and the Hardy–Weinberg equilibrium were investigated using a HWE-analysis software package (HWE-Analysis, Version 3.2 developed by Christoph Puers, Institute of Legal Medicine, University of Muenster, Germany).

3. Results and discussion

The allele frequencies and the statistical inferences for each of the investigated STR loci are shown in Table 1. The overall discrimination power was 0.999997 and the combined power of exclusion for the set of 10 STR loci reached 0.99991. Table 1 summarizes the testing results for the correspondence of genotype frequencies with the Hardy–Weinberg equilibrium proportions. No deviations from the Hardy–Weinberg equilibrium were observed ($p>0.05$ based on 5000 shuffling).

4. Conclusions

A reference database for the Romanian population has been established for a set of 10 STR loci. The results of this population study are very useful in current forensic practice with Romanian individuals in order to estimate the frequency of DNA profiles in casework as well as in paternity testing. The scientific data can also be of great interest for interpopulation comparisons.

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