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SHORT COMMUNICATION

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Genetic variation of the nine Profiler Plus loci in Russians

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Abstract This paper presents allele frequency distributions from a representative population sample of the Russian Federation for the nine loci (D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317 and D7S820) which constitute the commercially available Profiler Plus PCR amplification kit. DNA samples of 402 Russian individuals from 57 regions of the Russian Federation were amplified in a multiplex reaction with subsequent genotyping using an ABI Prism 377 DNA sequencer. The population data obtained for genotype and allele frequencies conformed to Hardy-Weinberg expectations (HWE).

Keywords STRs · DNA typing · Profiler Plus · Population studies · Russian Federation

Introduction

The polymorphisms of non-coding short repeat sequences of nuclear DNA (short tandem repeats, STRs) are widely used in legal medicine for the purposes of human identification. For more effective work on forensic biological material and acceleration of the genotyping procedure, multilocus AmpFLP systems based on simultaneous detection of polymorphisms for a number of STR loci are used [1, 2]. There are commercial kits on the market which are used for STR DNA typing in forensic casework and medicine. The AmpF/STR Profiler Plus PCR amplification kit is a commercial kit which enables co-amplification of the nine highly polymorphic STR loci D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, and D7S820. This multiplex is increasingly being used in forensic casework. While some of these STRs (e.g., vWA, FGA and D21S11) have already been studied and validated for forensic proposes, for Russia there is a lack of population and genetic data. In this paper, we used the AmpF/STR Profiler Plus kit for studying the genetic variability of the nine STR loci of this multiplex among a representative group of Russians living in 57 regions of the Russian Federation.

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Materials and methods

Blood samples from 402 unrelated Russian individuals living in 57 regions of the Russia (randomly sampled) were collected on FTA cards. DNA was extracted from the blood samples using the Chelex 100 method [3].

The PCR amplification of the nine STR loci D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, and D7S820 was performed using the AmpF/STR Profiler Plus amplification kit (PE Applied Biosystems) according to the manufacturer's instructions. Amplification was carried out in a DNA thermocycler GeneAmp PCR System 9700 (PE Applied Biosystems). Electrophoresis, detection of PCR products, and genotyping was carried out on the ABI Prism 377 DNA sequencer (PE Applied Biosystems) using the Genescan 3.1 and Genotyper 2.5 analysis software (PE Applied Biosystems). The population genetic parameters were estimated with the aid of Powerstat algorithms (Promega Corp.) [4, 5, 6].

Results and discussion

The data on allele frequencies for the AmpF/STR Profiler Plus loci for the Russian population samples studied are summarised in Table 1. All nine loci showed no significant deviation from Hardy-Weinberg expectations (Table 2). From the nine STR loci studied, locus D18S51 seems to be the most informative. The distribution of allele frequencies in the Russian population (our data) are confirmed by the similar data for Slovenian [7], Polish [8] and German [9] populations. The greatest distinctions between these populations were observed for loci D18S51 for alleles 12, 13 and 15.

Table 1 Allele frequencies of AmpF/STR Profiler Plus STR loci in Russians

Allele	Allele frequencies								
	vWA	D3S1358	FGA	D8S1179	D21S11	D18S51	D5S818	D13S317	D7S820
6	–	–	–	–	–	–	–	–	0.0012
7	–	–	–	–	–	–	0.0112	–	0.0087
8	–	–	–	0.0075	–	–	0.0012	0.1393	0.1928
9	–	–	–	0.0137	–	–	0.0522	0.0883	0.1480
10	–	–	–	0.0659	–	0.0050	0.0858	0.0684	0.2524
11	–	–	–	0.0734	–	0.0187	0.3496	0.3706	0.2040
12	–	–	–	0.1891	–	0.0958	0.3271	0.2040	0.1580
13	0.0025	0.0025	–	0.3234	–	0.1244	0.1642	0.0871	0.0299
14	0.0796	0.1169	–	0.2015	–	0.1368	0.0075	0.0423	0.0050
14.2	–	–	–	–	–	0.0012	–	–	–
15	0.0920	0.2537	–	0.1020	–	0.1704	0.0012	–	–
16	0.2127	0.2898	–	0.0160	–	0.1555	–	–	–
17	0.2836	0.1990	0.0025	0.0075	–	0.1343	–	–	–
18	0.2251	0.1269	0.0112	–	–	0.0759	–	–	–
19	0.0833	0.0100	0.0709	–	–	0.0410	–	–	–
20	0.0199	0.0012	0.1542	–	–	0.0174	–	–	–
21	0.0012	–	0.1629	–	–	0.0112	–	–	–
21.2	–	–	0.0012	–	–	–	–	–	–
22	–	–	0.1953	–	–	0.0075	–	–	–
22.2	–	–	0.0012	–	–	–	–	–	–
23	–	–	0.1493	–	–	0.0037	–	–	–
23.2	–	–	0.0025	–	–	–	–	–	–
24	–	–	0.1294	–	–	0.0012	–	–	–
25	–	–	0.0846	–	–	–	–	–	–
26	–	–	0.0311	–	0.0012	–	–	–	–
27	–	–	0.0037	–	0.0224	–	–	–	–
28	–	–	–	–	0.1443	–	–	–	–
29	–	–	–	–	0.2077	–	–	–	–
29.2	–	–	–	–	0.0012	–	–	–	–
30	–	–	–	–	0.2587	–	–	–	–
30.2	–	–	–	–	0.0572	–	–	–	–
31	–	–	–	–	0.0784	–	–	–	–
31.2	–	–	–	–	0.0759	–	–	–	–
32	–	–	–	–	0.0199	–	–	–	–
32.2	–	–	–	–	0.0871	–	–	–	–
33	–	–	–	–	0.0025	–	–	–	–
33.2	–	–	–	–	0.0423	–	–	–	–
34.2	–	–	–	–	0.0012	–	–	–	–

Table 2 The basic population characteristics of loci D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, and D7S820 for Russians

Statistical values	Locus								
	vWA	D3S1358	FGA	D8S1179	D21S11	D18S51	D5S818	D13S317	D7S820
Matching probability	0.073	0.081	0.038	0.068	0.042	0.030	0.113	0.078	0.064
Power of discrimination	0.927	0.919	0.962	0.932	0.958	0.970	0.887	0.922	0.936
PIC	0.773	0.748	0.843	0.772	0.826	0.864	0.690	0.752	0.782
Power of exclusion	0.648	0.527	0.726	0.610	0.667	0.696	0.454	0.555	0.547
Typical paternity index	2.87	2.09	3.72	2.58	3.05	3.35	1.76	2.23	2.18
Observed heterozygosity	0.826	0.761	0.866	0.806	0.836	0.851	0.716	0.776	0.771
Expected heterozygosity	0.803	0.783	0.860	0.799	0.845	0.878	0.735	0.781	0.811
χ^2-Value	2.54	13.01	10.49	0.40	0.51	37.01	5.90	6.75	10.53

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