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Variation of 15 Autosomal Microsatellite DNA Loci in the Russian Population

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Abstract—The allele frequencies of 15 autosomal STR loci (D3S1358, *vWA*, *FGA*, *TH01*, *TPOX*, *CSF1PO*, D5S818, D13S317, D7S820, D16S539, D2S1338, D8S1179, D21S11, D18S51, and D19S433) used in forensic medicine were determined for the Russian population of European Russia ($N = 176$). The power of discrimination (PD) and power of exclusion (PE) of the system of the 15 STR loci were 0.999 999 999 999 986 and 0.999 999 331 310 171 000, respectively. The allele and genotype frequency distributions in the Russian population corresponded to the Hardy–Weinberg equilibrium. The D2S1338, D18S51, D21S11, and *FGA* loci were identified as the most informative markers for the Russian population and proposed as a reference for forensic studies in the Russian Federation.

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Key words: autosomal microsatellite loci, human population, genetic diversity, DNA identification

INTRODUCTION

The variation of short tandem repeats (STRs), or microsatellites, dispersed through the whole human genome, is actively investigated in forensic labs for personal identification and determination of genetic relationships between individuals [1, 2]. Progress in studying the variation of autosomal microsatellite loci has become possible due to highly efficient multilocus DNA amplification systems, such as AmpFISTR SGM Plus and AmpFISTR Profiler. Thus, multilocus detection of autosomal microsatellites is now one of the most powerful tools in studies of genetic variation.

In spite of progress in creating databases of the allele and genotype frequency distributions of STR loci in various groups of the global population and individual ethnic groups, the body of information about polymorphic autosomal microsatellite loci in the ethnically differentiated population of the Russian Federation is still insufficient. The most part of the information obtained for the Russian population represents data on the allele frequency distributions of several loci used in forensic studies [3–5]. The use of a panel of nine STR loci (D3S1358, *vWA*, *FGA*, D8S1179, D21S11, D18S51, D5S818, D13S317, and D7S820) included into the AmpFISTR Profiler Plus system to study the genetic variation in ethnic Russians from different (European and Asian) regions of

the Russian Federation essentially increased the power of discrimination (PD) of indirect genetic personal identification [6]. However, the variation of autosomal STR loci in Russians from European Russia is still insufficiently characterized. In this work, we studied the variation of 15 microsatellite loci in 176 ethnic Russians from European Russia, using AmpFISTR SGM Plus and AmpFISTR Profiler multiplex PCR amplification systems.

EXPERIMENTAL

Sample. Genomic DNA was isolated from venous blood, using a standard technique, which included treatment with a detergent (1% SDS) and proteinase K (Sigma) and chloroform–phenol extraction [7]. The sample included 176 unrelated ethnic Russians from the southern part of European Russia: Stavropol ($N = 59$), Orel ($N = 72$), and Saratov ($N = 45$) regions.

Genotyping. STR loci were amplified using AmpFISTR SGM Plus and AmpFISTR Profiler multiplex systems as recommended by Applied Biosystems. The amplification products were electrophoretically separated using an ABI Prism 377 DNA sequencer (Applied Biosystems). Control DNA samples and allele standards were used for quality control. The size of the PCR products was determined using the GeneScan

Allele frequencies and other statistical parameters of 15 STR loci in the Russian population ($N = 352$ chromosomes)

Allele	D3S1358	VWA	FGA	TH01	TPOX	CSF1PO	D5S818	D13S317	D7S820	D16S539	D2S1338	D8S1179	D21S11	D18S51	D19S433
6	-	-	-	0.224	0.003	-	-	-	-	-	-	-	-	-	-
7	-	-	-	0.139	-	-	0.003	0.009	-	-	-	-	-	-	-
8	-	-	-	0.080	0.571	-	0.003	0.168	0.003	0.011	-	0.003	-	-	-
9	-	-	-	0.241	0.080	0.048	0.054	0.156	0.088	0.088	-	0.003	-	-	-
9.3	-	-	-	0.310	-	-	-	-	-	-	-	-	-	-	-
10	-	-	-	0.003	0.071	0.287	0.085	0.293	0.060	0.060	-	0.045	-	0.003	0.000
10.3	-	-	-	0.003	-	-	-	-	-	-	-	-	-	-	-
11	-	-	-	-	0.244	0.315	0.335	0.207	0.298	0.298	-	0.040	-	0.011	0.003
12	0.003	-	-	-	0.031	0.284	0.361	0.148	0.324	0.324	-	0.162	-	0.094	0.077
12.2	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.003
13	-	0.003	-	-	-	0.045	0.145	0.020	0.193	0.193	-	0.349	-	0.111	0.219
13.2	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.011
14	0.102	0.065	-	-	-	0.011	0.011	0.043	0.026	0.026	-	0.267	-	0.162	0.361
14.2	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.037
15	0.290	0.114	-	-	-	0.009	0.003	0.006	-	-	-	0.105	-	0.190	0.159
15.2	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.043
16	0.321	0.213	-	-	-	-	-	-	-	-	0.048	0.017	-	0.173	0.043
16.2	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.023
17	0.196	0.270	-	-	-	-	-	-	-	-	0.219	0.006	-	0.111	0.006
17.2	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.009
18	0.082	0.224	0.014	-	-	-	-	-	-	-	0.097	0.003	-	0.065	0.000
18.2	-	-	-	-	-	-	-	-	-	-	-	-	-	-	0.009
19	0.006	0.099	0.088	-	-	-	-	-	-	-	0.151	-	-	0.034	-
20	-	0.011	0.145	-	-	-	-	-	-	-	0.125	-	-	0.028	-
21	-	-	0.159	-	-	-	-	-	-	-	0.028	-	-	0.011	-
21.2	-	-	0.003	-	-	-	-	-	-	-	-	-	-	-	-
22	-	-	0.236	-	-	-	-	-	-	-	0.014	-	-	0.006	-
22.2	-	-	0.003	-	-	-	-	-	-	-	-	-	-	-	-

Table (Contd.)

Allele	D3S1358	VWA	FGA	TH01	TPOX	CSF1PO	D5S818	D13S317	D7S820	D16S539	D2S1338	D8S1179	D21S11	D18S51	D19S433
23	-	-	0.094	-	-	-	-	-	-	-	0.108	-	-	-	-
23.2	-	-	0.003	-	-	-	-	-	-	-	-	-	-	-	-
24	-	-	0.122	-	-	-	-	-	-	-	0.094	-	-	-	-
25	-	-	0.102	-	-	-	-	-	-	-	0.097	-	-	-	-
26	-	-	0.028	-	-	-	-	-	-	-	0.020	-	0.006	-	-
27	-	-	0.003	-	-	-	-	-	-	-	-	-	0.020	-	-
28	-	-	-	-	-	-	-	-	-	-	-	-	0.156	-	-
28.2	-	-	-	-	-	-	-	-	-	-	-	-	0.006	-	-
29	-	-	-	-	-	-	-	-	-	-	-	-	0.205	-	-
29.2	-	-	-	-	-	-	-	-	-	-	-	-	0.003	-	-
30	-	-	-	-	-	-	-	-	-	-	-	-	0.219	-	-
30.2	-	-	-	-	-	-	-	-	-	-	-	-	0.068	-	-
31	-	-	-	-	-	-	-	-	-	-	-	-	0.085	-	-
31.2	-	-	-	-	-	-	-	-	-	-	-	-	0.080	-	-
32	-	-	-	-	-	-	-	-	-	-	-	-	0.020	-	-
32.2	-	-	-	-	-	-	-	-	-	-	-	-	0.085	-	-
33.2	-	-	-	-	-	-	-	-	-	-	-	-	0.045	-	-
34.2	-	-	-	-	-	-	-	-	-	-	-	-	0.003	-	-
MP	0.098	0.067	0.041	0.092	0.205	0.127	0.122	0.070	0.072	0.100	0.035	0.095	0.039	0.039	0.073
PD	0.902	0.933	0.959	0.908	0.795	0.873	0.878	0.930	0.928	0.900	0.965	0.905	0.961	0.961	0.927
PIC	0.72	0.78	0.84	0.73	0.55	0.68	0.68	0.77	0.77	0.72	0.86	0.73	0.84	0.85	0.76
PE	0.481	0.570	0.745	0.570	0.287	0.520	0.500	0.655	0.560	0.560	0.779	0.622	0.699	0.802	0.520
PI	1.87	2.32	4.00	2.32	1.24	2.05	1.96	2.93	2.26	2.26	4.63	2.67	3.38	5.18	2.05
H_0	0.7330	0.7841	0.8750	0.7841	0.5966	0.7557	0.7443	0.8296	0.7784	0.7784	0.8921	0.8125	0.8523	0.9034	0.7557
H_e	0.7623	0.8080	0.8581	0.7719	0.6036	0.7350	0.7282	0.8001	0.7988	0.7624	0.8735	0.7719	0.8608	0.8701	0.7874
P (HW)	0.9245	0.6205	0.9606	0.5845	0.8428	0.3056	0.6166	0.8857	0.9819	0.8892	0.3944	0.8887	0.1574	0.2273	0.3588

Note: MP, genotype match probability; PD, power of discrimination; PIC, polymorphism information content; PE, power of exclusion; PI, paternity index; H_0 , observed heterozygosity; H_e , expected heterozygosity; P (HW), probability of deviation from the Hardy-Weinberg equilibrium.

(v. 3.1) and Genotyper (v. 2.0) software (Applied Biosystems).

Statistical analysis of the data was carried out using the Arlequin software package (v. 2.000) [8] and the PowerTyper Excel spreadsheet program (Promega) [9].

RESULTS AND DISCUSSION

The allele frequencies of 15 STR loci (D3S1358, *vWA*, *FGA*, *TH01*, *TPOX*, *CSFIPO*, D5S818, D13S317, D7S820, D16S539, D2S1338, D8S1179, D21S11, D18S51, and D19S433) assayed in 176 Russians are shown in the table. The observed genotype frequencies corresponded to the Hardy–Weinberg distribution. PD of the multiplex DNA amplification systems was very high. For example, PD and the power of exclusion (PE) of the locus panel in the Russian population were 0.999 999 999 999 986 and 0.999 999 331 310 171 000, respectively. Four of the 15 loci (D2S1338, D18S51, D21S11, and *FGA*) were the most informative for genetic personal identification ($PIC > 0.8$, see the table). Comparative analysis of our data and earlier results of typing nine STR loci (D3S1358, *vWA*, *FGA*, D8S1179, D21S11, D18S51, D5S818, D13S317, and D7S820) in the Russian population [6] showed a similar character of allele frequency distribution. Comparison of the allele frequency distribution for the STR loci from the AmpFISTR SGM Plus panel in the Russian and other Slavic populations (Poles [10], Slovenes [11], Serbs [12], and Bosnians [13]) revealed no reliable distinctions in F_{ST} . Compared with the above Slavic ethnic groups, Russians were characterized by a higher frequency of allele D3S1358*16 and lower frequencies of alleles D13S317*12, *vWA**14, D3S1358*18, and *FGA**21. At the same time, Russians and Poles are similar in the distribution of alleles 12, 13, and 15 of D18S51. Moreover, there were no reliable distinctions between Russians and Poles regarding the allele frequency distributions of the STR loci included in both PCR amplification systems [11].

Thus, the data obtained in our work show that analysis of the variation of the autosomal STR loci included in the AmpFISTR SGM Plus and AmpFISTR Profiler systems is promising for personal identification in the Russian population. In view of a sufficiently great number of the STR loci examined in Russians from the southern regions of European Russia, our results make an important contribution to the creation of a reference database of molecular data, required for further improvement of the statistical basis of forensic studies in the Russian Federation.

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