

Towards Russian reference population data on STR loci

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Abstract. Summary of allele counts for 10 STR loci: CSF1PO, D7S820, D13S317, F13A01, F13B, FESFPS, LPL, TH01, TPOX and vWA obtained by the seven laboratories in a countrywide Russian Caucasians is proposed as a reference population data set. © 2003 Elsevier B.V. All rights reserved.

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1. Introduction

At least three Ministries (Ministry of Health, Ministry of Defense and Ministry of Internal Affairs) in Russia have specialized forensic genetics laboratories in several Federal districts. Moreover, some forensic DNA markers are used in several academic institutes for the studies in ethnogenomics or genogeography, human molecular medicine and genetics [1]. Data accumulated by these laboratories seem to be suitable for creating Russian reference population data bank (see, e.g., [2–5]).

2. Population

Several hundred unrelated Caucasians living in about 60 cities and regions (federal cities Saint Petersburg and Moscow, and main federal districts: Northwestern, Central, Northern Caucasus, Volga, Ural, Siberia and Far East) were examined.

3. Extraction

Chelex® 100, phenol-chloroform and/or Expert 1 [6] extraction protocols adapted for, and widely used in forensic studies.

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4. PCR and typing

GenePrint® STR Systems, GenePrint® Fluorescent STR Systems (Promega) and AmpF/STR Profiler Plus amplification kit (PE Applied Biosystems) according to the manufacturer's instructions.

5. Data analysis

To test Hardy–Weinberg equilibrium and homogeneity between samples the PC programs which provide exact randomization procedures based on different statistics (probability, chi-square, F_{IS} —fixation indices, etc.) were used (see, e.g., lists in Refs. [3,5]). To construct 95% credible (Bayesian) limits for coancestry coefficient F_{ST} (the correlation of two genes sampled from distinct individuals within the same subpopulation, relative to the total population), the FSTMET routine was applied (<http://www.rdg.ac.uk/statistics/genetics/software.html>, <http://www.reading.ac.uk/~snsbalng/>) (see Refs. [7–10]).

6. Access to the data

Detailed results as well as original individual genotype data in form of input files suitable for the use with different software for the population genetic analysis (Arlequin, FSTAT, FSTMET, GDA, GENEPop, GENETIX, HWE, HWMET, POPGENE, Popula-

Table 1
Allele counts for 10 STR loci in combined samples of unrelated Russian Caucasians

Allele	Locus									
	CSF1PO	D7S820	D13S317	F13A01	F13B	FESFPS	LPL	TH01	TPOX	vWA
3,2				15						
4				35						
5				67				1		
6		1		132	59			286	4	
7	1	12	1	137	11		2	246	3	
8	0	256	195	4	180	24	2	158	520	
9	41	191	121	0	147	7	32	287	70	
9,3	–	–	–	–	–	–	–	413	–	
10	194	349	84	0	292	333	327	37	49	
11	201	280	504	0	6	581	175	2	195	
12	210	219	284	1	3	228	127		23	
13	59	44	109	0		68	19			2
14	9	6	60	5		3				136
15	1			14						177
16				2						322
17										501
18										366
19										140
20										31
21										1
Total allele count	716	1358	1358	412	698	1244	684	1430	864	1676
Lab No. ^a	1,2,5	3,4,5,7	3,4,5,7	1,2	1,6	1,2,5,6	1,3,4	1,3,4, 5,6	1,2,5	1,2,3, 4,5
P^b	0.048	0.313	0.427	0.065	0.350	0.463	0.537	0.117	0.069	0.775
95% CL for F_{ST}^c	0.0006	0.0002	0.002	0.004	0.0003	0.0003	0.0003	0.0004	0.0005	0.0002
	0.019	0.0077	0.012	0.017	0.019	0.0056	0.022	0.020	0.014	0.004

^a Number of the lab as shown in the authors' affiliation above.

^b P -value for the inter-laboratory homogeneity.

^c Credible (Bayesian) limits for the posterior distribution of coancestry coefficient.

tion, PowerMarker, PowerStats, TFPGA, etc.) are available to interested users upon request from the authors SAG@cards.lanck.net or Nikita@NH8333.spb.edu.

7. Results

In each data set presented by the participated laboratories, no significant deviation from Hardy–Weinberg equilibrium was detected. Genotype and allele distributions obtained by the laboratories appeared to be in good agreement (statistically homogeneous) and so could be combined into single reference sample as shown below (Table 1). Evaluated values for coancestry coefficient with the upper 95% credible limit about 0.02 are compatible with those reported for other Caucasians over the world [7,8,10].

8. Other remarks

We insist positively on publication of the absolute allele frequencies so that any user could to calculate relative values with a precision preferred.

Russian Federation consists of seven federal districts that are divided into 89 “subjects” which are: 21 autonomous republics, 1 autonomous oblast, 10 autonomous districts, six regions and 49 oblasts and two federal cities where about 130 different nations are living (see, e.g., http://www.wikipedia.org/wiki/Districts_in_Russia). So further comprehensive study involving samples from different nationalities and ethnic groups is required.

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References

- [1] S.A. Limborska, et al., *Ethnogenomics and Gene Geography of Peoples from Eastern Europe*, Nauka, Moscow, 2002, 261 pp.
- [2] V.P. Pushkarev, P.I. Novikov, The use of capillary electrophoresis for the study of D13S317 and D7S820 microsatellites in Caucasian population of the Ural region of Russia, *Forensic Med. Expert.* 45 (6) (2002) 31–35 (in Russia).
- [3] A.G. Smolyanitsky, et al., Statistical pattern analysis of D1S80 alleles in Northwestern Russians and worldwide database using COLLAPSE software, in: B. Brinkman, A. Carracedo (Eds.), *Progress in Forensic Genetics*, vol. 9, Elsevier, Amsterdam, 2003, pp. 665–671.
- [4] I.V. Kornienko, et al., Genetic variation of the nine Profiler Plus loci in Russians, *Int. J. Leg. Med.* 116 (2002) 309–311.
- [5] A.G. Smolyanitsky, et al., Polymorphism of LDLR, GYPA, HBGG, D7S8, GC, HLA-DQA1, Ig-JH, D17S30, ApoB and D1S80 loci in northwestern Russians, *Forensic Sci. Int.* (2003) 100–103.
- [6] A. Smolyanitsky, et al., Applications of Expert 1 kit for genomic DNA isolation from forensic biological samples: muscle, blood stains, sperm stains and single hairs, *Med. Leg. Baltica* 7 (1996) 58–61.
- [7] D.J. Balding, et al., Population genetics of STR loci in Caucasians, *Int. J. Leg. Med.* 108 (1996) 300–305.
- [8] D.J. Balding, R.A. Nichols, Significant genetic correlations among Caucasians at forensic DNA loci, *Heredity* 78 (1997) 583–589.
- [9] W. Lee, et al., Statistical analysis for estimating heterogeneity of the Korean population in DNA typing using STR loci, *Int. J. Leg. Med.* 116 (2002) 153–160.
- [10] B.S. Weir, W.G. Hill, Estimating F-statistics, *Annu. Rev. Genet.* 36 (2002) 721–750.