Journal of
APPLIED
BIOMEDICINE

J. Appl. Biomed.
3: 129–131, 2005
ISSN 1214-0287

ORIGINAL ARTICLE

Population genetics of 11 nuclear and 1 mitochondrial short tandem repeat loci in a population of South Bohemia, Czech Republic

Tomáš Vaněček [1, 3 #], Pavel Trubač [2, 3, #], František Vorel [2], Miroslav Šíp [4]

[1] Sikl`s Department of Pathology, Charles University Hospital, Plzeň, Czech Republic
[2] Department of Forensic Medicine, University Hospital, České Budějovice, Czech Republic
[3] Faculty of Biological Science, University of South Bohemia, České Budějovice, Czech Republic
[4] Faculty of Health and Social Studies, University of South Bohemia, České Budějovice, Czech Republic
[#]These authors contributed equally to the work.

Received 28th February 2005.
Revised 12th April 2005.
Published online 10th June 2005.

Summary
A population study on 11 nuclear and 1 mitochondrial short tandem repeats (STR) (CSF1PO, TPOX, THO1, F13A01, FESFPS, vWA, D16S539, D7S820, D13S317, LPL, F13B and mitochondrial $(CA)_n$ repeat (mtCA)) was performed, and allele frequencies together with other forensic genetics characteristics, were estimated from a sample of 111 – 253 unrelated individuals living in the area of South Bohemia, Czech Republic. All loci, except F13B, meet Hardy-Weinberg expectations. Significant differences of allele frequencies were found in three (vWA, D16S539 and F13B) or two loci (vWA, D16S539) when compared with reference or known Czech data, respectively. An allele frequencies comparison of the mitochondrial marker shows significant differences only in the case of the African population.

Keywords : short tandem repeat – population data – GenePrint STR Kit – mitochondrial CA repeat polymorphism –Czech Republic

INTRODUCTION

The PCR amplification of STR markers is at present the most common tool in the DNA analysis of individuals and traces in forensic genetics practice (Gill 2002). The population specific frequency of each of the alleles of the STR loci are known to receive correct statistical results of forensic genetic typing (Wall et al. 1993). In this article, we present, in a format recommended for the publication of such genetic data (Lincoln and Carracedo 2000), allele frequencies of 11 nuclear and 1 mitochondrial short tandem repeat loci of the South Bohemian population. This will be helpful in forensic genetics practice, but also the information represents a contribution to the general knowledge of European population genetics.

✉ Miroslav Šíp, Faculty of Health and Social Studies, University of South Bohemia, České Budějovice, Czech Republic
sip@zsf.jcu.cz
☎ +420 386 352 621

Table 1. **Allelic frequencies of GenePrint STR systems and mitochondrial STR marker (mtCA) of the South Bohemian population**

allele	CSF1PO	TPOX	THO1	F13A01	FESFPS	VWA	D16S539	D7S820	D13S317	LPL	F13B	mtCA
3.2	-	-	-	0.100	-	-	-	-	-	-	-	-
4	-	-	-	0.047	-	-	-	-	-	-	-	0.090
5	-	-	0.002	0.174	-	-	-	-	-	-	-	0.820
6	-	-	0.228	0.291	-	-	-	-	-	-	0.096	0.063
7	-	-	0.161	0.338	-	-	-	0.016	-	-	-	0.027
8	-	0.537	0.089	-	0.017	-	0.012	0.156	0.135	0.011	0.202	-
9	0.024	0.104	0.220	-	0.002	-	0.126	0.154	0.073	0.032	0.241	-
9.3	-	-	0.289	-	-	-	-	-	-	-	-	-
10	0.256	0.058	0.011	-	0.260	-	0.051	0.251	0.052	0.439	0.443	-
11	0.300	0.271	-	-	0.444	-	0.257	0.231	0.349	0.245	0.011	-
12	0.326	0.030	-	0.007	0.221	-	0.360	0.138	0.262	0.252	0.007	-
13	0.066	-	-	0.005	0.051	-	0.156	0.036	0.097	0.022	-	-
14	0.026	-	-	0.015	0.005	0.078	0.036	0.018	0.028	-	-	-
15	0.002	-	-	0.017	-	0.088	0.002	-	0.004	-	-	-
16	-	-	-	0.005	-	0.228	-	-	-	-	-	-
17	-	-	-	-	-	0.260	-	-	-	-	-	-
18	-	-	-	-	-	0.245	-	-	-	-	-	-
19	-	-	-	-	-	0.098	-	-	-	-	-	-
20	-	-	-	-	-	0.002	-	-	-	-	-	-
N	227	231	230	201	204	204	253	253	252	139	141	111
GD	-	-	-	-	-	-	-	-	-	-	-	0.318
RMP	-	-	-	-	-	-	-	-	-	-	-	0.685
hO	0.767	0.623	0.722	0.766	0.696	0.814	0.755	0.798	0.750	0.662	0.610	-
hE	0.733	0.624	0.782	0.758	0.684	0.797	0.760	0.814	0.773	0.683	0.695	-
PD	0.871	0.800	0.922	0.901	0.835	0.923	0.900	0.939	0.916	0.837	0.851	-
PE	0.538	0.320	0.463	0.538	0.422	0.625	0.518	0.596	0.510	0.372	0.303	-
PI	2.142	1.328	1.797	2.138	1.645	2.684	2.040	2.480	2.000	1.479	1.282	-
P	0.687	0.444	0.185	0.354	0.176	0.699	0.275	0.580	0.732	0.111	0.0005	-

N - number of samples, hO - observed heterozygosity, hE - expected heterozygosity, PD - power of discrimination, PE - power of exclusion, PI - typical paternity index, P - Fisher`s exact test for Hardy-Weinberg equilibrium, based on 5000 iterations (Raymond and Rousset 1995), GD - genetic diversity (Tajima 1989), RMP - random match probability (Stoneking *et al.* 1991).

MATERIAL AND METHODS

*Population***:** Unrelated individuals living in the region of South Bohemia, Czech Republic.
*DNA extraction***:** DNA was extracted by QIAamp DNA Blood Kit (Qiagen, Germany).
*PCR***:** The amplification of nuclear STR markers was performed using the GenePrint STR System Amplification Kit according to the manufacturer`s instructions (Promega, Germany). The amplification of the mitochondrial STR marker (mtCA) (3` area of the mitochondrial control region) (Szibor et al. 1997) was carried out as described in Vaněček et al. (2004).

*Detection***:** In the case of nuclear markers, PCR products and reference sequenced allelic ladders were separated by 6% denaturing vertical polyacrylamide gel electrophoresis and visualized by silver staining according to Budowle et al. (1991). The typing of the mitochondrial marker was performed by sequencing as described in Vaněček et al. (2004).
*Data analysis***:** The nuclear markers data were analysed using GenePop (Raymond and Rousset 1995a) and PowerStats software (http://www.promega.com/geneticidtools/powerstats/Default.htm). The mitochondrial marker data

characteristics were counted according to Tajima (1989) and Stoneking et al. (1991). Our nuclear STR data were compared with reference to the Caucasian sample population (www.promega.com/techserv/apps/hmnid/reference information/popstat/custstat_Allelefreq.htm). Mitochondrial marker data was compared with data published by Szibor et al. (1997). Statistical comparisons based on the Fisher exact test of RxC contingency tables through the use of the Metropolis algorithm (Raymond and Rousset 1995b) were carried out by using software kindly supplied by Mark P. Miller, the significance level $2\alpha=0.05$ was used.

RESULTS AND DISCUSSION

The results of our typing of 11 nuclear and one mitochondrial STR markers are shown in Table 1.

All loci, except F13B (statistically significant), meet Hardy-Weinberg expectations. An allele frequency comparison between our data and the reference data reveals no significant differences, except vWA, D16S539 and F13B (statistically significant) loci. Additionally, loci vWA and D16S539 differ significantly also from known Czech population data (Vaněk et al. 2001) – the data of the Czech population for F13B are not available. The MtCA marker data show no significant difference from any other European Caucasian population. On the other hand, mitochondrial data do reveal significant differences when compared to the African population (Szibor et al. 1997). The calculated random match probability of the mitochondrial marker, an equivalent for matching probability (1-PD), attains a value of 0.685 which is more than for most nuclear markers, but this disadvantage is balanced, for example, by the possibility of amplification of mtDNA from highly degraded material (Seo et al. 2000).

In our study we determined allele frequencies and other characteristics of 12 STR markers of the South Bohemian population. These data are more relevant for computation of "local specific" forensic genetic parameters than reference data and enable us to obtain more accurate results samples typing in future.

ACKNOWLEDGEMENT

This study was partially supported by grant No. 350/2000 of the Ministry of Education of the Czech Republic.

REFERENCES

Budowle B., Chakraborty R., Giusti A.M., Eisenberg A.J., Allen R.C.: Analysis of the VNTR locus D1S80 by the PCR followed by high-resolution PAGE. Am. J. Hum. Genet. 48:137–144, 1991.

Gill P.: Role of short tandem repeat DNA in forensic casework in the UK – past, present and future perspectives. Biotechniques 32:366–385, 2002.

Lincoln P., Carracedo A.: Publication of population data of human polymorphisms. Forensic Sci. Int. 110:3–5, 2000.

Raymond M.L., Rousset F.: GENEPOP (version 1.2): population genetics software for exact tests and ecumenicism. J. Heredity 86:248–249, 1995a.

Raymond M.L., Rousset F.: An exact test for population differentiation. Evolution 49:1280-1283, 1995b

Seo Y., Uchiyama T., Shimizu K., Takahama K.: Identification of remains by sequencing of mitochondrial DNA control region. Am. J. Forensic Med. Pathol. 21:138–143, 2000.

Stoneking M., Hedgecock D., Higuchi R.G., Vigilant L., Erlich H.A.: Population variation of human mtDNA control region sequences detected by enzymatic amplification and sequence-specific oligonucleotide probes. Am. J. Hum. Genet. 48:370–382, 1991.

Szibor R. et al.: Mitochondrial D-loop 3' (CA)n repeat polymorphism: optimization of analysis and population data. Electrophoresis 18:2857–2860, 1997.

Tajima F.: Statistical method for testing the neutral mutation hypothesis by DNA polymorphism. Genetics 123:585–595, 198

Vaněček T., Vorel F., Šíp M.: Mitochondrial DNA D-loop hypervariable regions: Czech population data. Int. J. Legal Med. 118:14–18, 2004.

Vaněk D., Hradil R., Budowle B.: Czech population data on 10 short tandem repeat loci of SGM Plus STR system kit using DNA purified in FTA cards. Forensic Sci. Int. 119:107–108, 2001.

Wall W.J., Williamson R., Petrou M., Papaioannou D., Parkin B.H.: Variation of short tandem repeats within and between populations. Hum. Mol. Genet. 2:1023–1029, 1993.