

SHORT COMMUNICATION

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Population genetics of the STRs vWA, D3S1358 and FGA in the Pomerania-Kujawy region of Poland

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Abstract This paper presents the results of a Polish population study ($n = 210$) for the three STR loci vWA, D3S1358 and FGA analysed using the multiplex PCR system AmpflSTR Blue. The allele distributions were in accordance with Hardy-Weinberg expectations. The combined mean exclusion chance, mean paternity index and power of discrimination for the three loci were MEC = 0.96055, MPI = 127.1295 and PD = 0.99986. This demonstrates that these systems are valuable tools for forensic identification and paternity testing.

Key words Short tandem repeats (STRs) · DNA · Multiplex PCR · Population genetics · Poland

Introduction

Highly polymorphic STR loci are very useful in human identification [1, 2] and many systems have been investigated using PCR [3–10]. Prior to the introduction of a new DNA typing method to forensic casework, a study on the allele frequencies and genotype distribution in the population is required.

Materials and methods

Blood samples were obtained from healthy unrelated individuals from the Pomerania-Kujawy Region of Poland. DNA was extracted from blood by the salting out procedure [11].

For DNA amplification and typing the AmpflSTR Blue (Perkin Elmer/ABD, Foster City, Calif.) multiplex STR system was used which allows co-amplification of the three STR loci D3S1358, vWA and FGA. DNA samples (1–2.5 ng) were amplified according to the manufacturers' instructions. Amplified fragments were resolved by electrophoresis in a 5% denaturing poly-

acrylamide gel using the ABI Prism 377 DNA sequencer. The fluorescent ladder CXR 60-400 bases (Promega) was included in each lane as an internal size standard. Allele size determination and genotyping were performed using ABI Prism GeneScan Analysis 2.1 and ABI Prism Genotyper 2.0 software by comparison of amplified fragments with internal size standards and allelic ladders.

Allele frequencies were calculated for each STR locus, the number of heterozygotes was determined and the heterozygosity index (HI) was established. Conformation to Hardy-Weinberg equilibrium was tested by χ^2 -analysis and the exact test using the SPSS statistical package [12]. The mean exclusion chance (MEC), mean paternity index (MPI) and power of discrimination (PD) were calculated for single loci and as a multiplex [12, 13].

Results and discussion

The allele frequencies for vWA, D3S1358 and FGA loci are shown in Table 1. Statistical parameters of forensic importance for the loci analysed are shown in Table 2.

Table 1 Allele frequencies for vWA, D3S1358 and FGA loci in a sample of 210 Caucasians from the Pomerania-Kujawy region of Poland

vWA		D3S1358		FGA	
Allele	Frequency	Allele	Frequency	Allele	Frequency
13	0.0048	13	0.0024	17	0.0024
14	0.1048	14	0.1310	18	0.0119
15	0.1048	15	0.2714	19	0.1143
16	0.1833	16	0.2381	20	0.1381
17	0.2857	17	0.1762	21	0.1238
18	0.1929	18	0.1714	21.2	0.0071
19	0.1167	19	0.0095	22	0.1619
20	0.0071			22.2	0.0095
				23	0.1143
				23.2	0.0071
				24	0.1143
				24.2	0.0024
				25	0.1595
				26	0.0238
				27	0.0095

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Table 2 Statistical parameters of forensic importance for the three STR loci vWA, D3S1358 and FGA in a Caucasian population from Poland

Locus	H _{obs}	H _{exp}	PD	MEC	MPI	df	χ ²	p	exact p
vWA	0.77143	0.81193	0.93886	0.62847	4.2970	22	16.422	0.794	0.784
D3S1358	0.86190	0.79196	0.92420	0.58622	3.8020	15	10.340	0.798	0.792
FGA	0.89524	0.87375	0.97057	0.74336	7.7816	18	23.209	0.183	0.172
AmpFISTR Blue	–	–	0.99986	0.96055	127.1295	–	–	–	–

H_{obs} – observed heterozygosity, H_{exp} – expected heterozygosity, PD – power of discrimination, MEC – mean exclusion chance, MPI – mean paternity index, df – degrees of freedom

The population tested in this study did not show any significant deviation from Hardy-Weinberg equilibrium ($p > 0.05$) for all loci.

In the FGA locus we observed four interalleles (21.2, 22.2, 23.2, 24.2) and one allele with 17 repeats. Data obtained in analysis of FGA locus in the Polish population are similar to those presented by Rolf et al. [14] for a German population.

The combined values of mean exclusion chance, mean paternity index and power of discrimination for the AmpFISTR Blue System in Polish population are similar to those obtained by Wallin et al. [15] for American Caucasians in a validation study of AmpFISTR Blue PCR amplification kit. These data confirm the usefulness of the AmpFISTR Blue multiplex system in both paternity testing and routine forensic casework.

References

1. Edwards A, Civitello A, Hammond HA, Caskey CT (1991) DNA typing and genetic mapping with trimeric and tetrameric tandem repeats. *Am J Hum Genet* 49:746–756
2. Sprecher CJ, Puers C, Lins AM, Schumm JW (1996) General approach to analysis of polymorphic short tandem repeat loci. *Biotechniques* 20(2):266–276
3. Kimpton C, Fisher D, Watson S, Adams M, Urquhart A, Lygo J, Gill P (1994) Evaluation of an automated DNA profiling system employing multiplex amplification of four tetrameric STR loci. *Int J Legal Med* 106:302–311
4. Sparkes R, Kimpton C, Watson S, Oldroyd N, Clayton T, Barnett L, Arnold J, Thompson C, Hale R, Chapman J, Urquhart A, Gill P (1996) The validation of a 7-locus multiplex STR test for use in forensic casework. (I) Mixtures, ageing, degradation and species studies. *Int J Legal Med* 109:186–194
5. Meyer E, Wiegand P, Brinkmann B (1995) Phenotype differences of STRs in 7 human populations. *Int J Legal Med* 107:314–322
6. Takeshita H, Meyer E, Brinkmann B (1997) The STR loci HumTPO and HumLPL: population genetic data in eight populations. *Int J Legal Med* 110:331–333
7. Sparkes R, Kimpton C, Gilbard S, Carne P, Andersen J, Oldroyd N, Thomas D, Urquhart A, Gill P (1996) The validation of a 7-locus multiplex STR test for use in forensic casework. (II) Artefacts, casework studies and success rates. *Int J Legal Med* 109:195–204
8. Lins AM, Sprecher CJ, Puers C, Schumm JW (1996) Multiplex sets for the amplification of polymorphic short tandem repeat loci – silver stain and fluorescence detection. *Biotechniques* 20:882–889
9. Miścicka-Śliwka D, Grzybowski T (1997) High microvariation sequence polymorphism at short tandem repeat loci: human beta-actin related pseudogene as an example. *Electrophoresis* 18:1613–1619
10. Miścicka-Śliwka D, Grzybowski T, Woźniak M (1997) Optimization of a hexaplex PCR amplification from short tandem repeats (STR) and amelogenin (AMG) loci. *Electrophoresis* 18:1627–1632
11. Miller SA, Dykes DD, Polesky HF (1988) A simple salting out procedure for extracting DNA from human nucleated cells. *Nucleic Acids Res* 16(3):1215
12. Weir BS (1996) Genetic data analysis II. Sinauer Associates, Sunderland, Mass, pp 92–112 and pp 209–211
13. National Research Council Report II. The evaluation of forensic DNA evidence (1996) National Academy Press, Washington DC, pp 96–97
14. Rolf B, Waterkamp K, Huhne J (1998) Allele frequency data for the FGA locus in eight populations. *Int J Legal Med* 111:55–56
15. Wallin JM, Buoncristiani MR, Lazaruk KD, Fildes N, Holt CL, Walsh PS (1998) TWGDAM validation of the AmpFISTR blue PCR amplification kit for forensic casework analysis. *J Forensic Sci* 43:854–870