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Population genetic analysis in a German population from Saxony-Anhalt using the Powerplex[™] 16 system

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Abstract. The use of polymorphic short tandem repeats (STRs) has become important in genetics applications, such as gene mapping, identification and paternity. In forensics, STRs are mainly used for paternity testing and personal identification. In this study, we present the results of a survey aimed at investigating the allele and genotype frequency distribution of 15 loci amplified by the GenePrint[®] PowerPlex[™] 16 system (Promega) in Saxony-Anhalt. DNA was isolated from blood and saliva samples, randomly selected from routine casework. More than 200 unrelated individuals were included in the database. Amplification products were analyzed by capillary electrophoresis using the ABI 310[®] Genetic Analyzer (Applied Biosystems). Statistical analysis was carried out using various statistical methods (Hardy-Weinberg-Equilibrium, Mean Exclusion Power, Discrimination Power, etc.) to determine allele frequencies and other population parameters of interest. © 2003 Elsevier B.V. All rights reserved.

Keywords: Population study; PowerPlex[™] 16; German; STRs

1. Introduction

The use of polymorphic short tandem repeats (STRs) has become important in genetics applications, such as gene mapping, identification and paternity. In forensics, STRs are mainly used for paternity testing and personal identification.

In this study, we present the results of a survey aimed at investigating the allele and genotype frequency distribution of 15 loci amplified by the GenePrint[®] PowerPlexTM 16 system (Promega) in Saxony-Anhalt.

2. Material and methods

Either blood samples or buccal swabs were taken from 207 Germans in Saxony-Anhalt (Fig. 1) involved in criminal and paternity cases. DNA was isolated using an alkaline lysis extraction protocol [1].

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Fig. 1. German population sample from Saxony-Anhalt.

Two milliliters of the extracts was amplified following the protocols described in the GenePrint[®] PowerPlexTM kit 16 manual with two modifications: No quantification of the DNA in the extracts was performed and the PCR volume was 12.5 μ l.

Amplification products were analyzed by capillary electrophoresis using the ABI 310[®] Genetic Analyzer according to the manufacturer's recommendations (Applied Biosystems).

Forensically relevant statistical analysis like Power of Discrimination or Power of Exclusion were calculated using the Powerstats 1.2 worksheet (Promega). Exact tests for checking the Hardy–Weinberg expectations were performed using the GENEPOP software version 1.2 [2].

3. Results and discussion

All loci met Hardy–Weinberg expectations. A part of the samples was typed using the AmpFLSTR[®] SGM Plus[®] Kit, which shares eight loci with the GenePrint[®] PowerPlex[™] kit. The loci typed in both kits did not reveal any discrepancies. The results for these loci have been reported earlier [3]. The results for the remaining seven loci and parameters of forensic interest are given in Table 1.

Table	1

Allelic frequencies and forensic efficiency parameters of 7 STR loci in 207 unrelated Germans

	Penta D	TPOX	Penta E	D5S818	D13S317	D7S820	CSF1P0
Forensic							
Matching probability	0.064	0.201	0.025	0.124	0.083	0.068	0.117
Expressed as 1 in.	15.68	4.98	40.16	8.07	12.00	14.79	8.54
Power of discrimination	0.936	0.799	0.975	0.876	0.917	0.932	0.883
PIC	0.788	0.575	0.880	0.667	0.738	0.787	0.695
Paternity							
Power of exclusion	0.603	0.307	0.714	0.430	0.501	0.752	0.517
Typical paternity index	2.524	1.293	3.569	1.674	1.959	4.121	2.035
Allele frequencies							
Homozygotes	0.198	0.387	0.140	0.299	0.255	0.121	0.246
Heterozygotes	0.802	0.613	0.860	0.701	0.745	0.879	0.754
	Allele	Frequency					
	5	_	_	0.101	_	_	_
	6	_	_	_	_	_	_
	7	0.007	_	0.169	_	_	0.027
	8	0.014	0.523	0.010	0.002	0.142	0.161
	9	0.213	0.084	0.007	0.050	0.082	0.176
	10	0.089	0.059	0.104	0.068	0.044	0.251
	11	0.155	0.290	0.109	0.384	0.347	0.215
	12	0.263	0.044	0.179	0.315	0.270	0.134
	13	0.188	-	0.080	0.174	0.079	0.033
	14	0.060	-	0.051	0.004	0.031	0.002
	15	0.010	_	0.051	0.002	0.004	_
	16	_	-	0.056	_	-	-
	17	_	-	0.034	_	-	-
	18	_	-	0.029	_	-	-
	19	_	-	0.010	_	-	-
	20	_	_	0.007	_	-	-
	21	_	-	-	_	-	-
	22	_	_	0.002	-	-	-
	23	_	_	-	-	-	-
	24	_	_	0.002	-	-	-
	26	-	_	-	-	-	-

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