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Flemish population genetic analysis using 15 STRs of the Identifiler® kit

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Abstract. Allelic frequencies for the short tandem repeat systems CSF1PO, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, D21S11, vWA, FGA, TH01, TPOX, D2S1338 and D19S433 were determined in a Flemish population sample of 231 individuals, using the Identifiler kit (Applied Biosystems). No deviations from Hardy–Weinberg equilibrium were observed. Combined, the 15 loci yield a matching probability of 1 in 111×10^{12} and a power of exclusion of 99.999995%.

Keywords: STR polymorphisms; Population genetics; Forensic genetics; Flemish

1. Introduction

The aim of this study was to establish a database of the Flemish, i.e., the Dutch speaking population of the northern half of Belgium. We therefore applied the AmpFlSTR Identifiler PCR Amplification (Applied Biosystems) kit, that co-amplifies the 13 Combined DNA Index System (CODIS) STR loci (CSF1PO, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, D21S11, vWA, FGA, TH01, TPOX) and in addition the two tetrameric markers D2S1338 and D19S433, as well as the amelogenin locus for gender identification. Here we present the allelic frequencies and parameters of forensic efficiency in a sample of 231 unrelated Flemish individuals.

2. Material and methods

Buccal swabs were collected from 231 unrelated Flemish individuals, representing the mother (115 females) or alleged father (116 males) from paternity cases. DNA was

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extracted using the Qiamp DNA kit (Qiagen). PCR amplification and subsequent capillary electrophoresis were performed according to the manufacturer's manual, on the PE 9700 thermal cycler (Applied Biosystems) and the ABI 3100 Genetic Analyzer (Applied Biosystems) respectively. Alleles were named according to the recommendations of the DNA Commission of the International Society for Forensic Genetics [1]. Allelic frequencies were estimated by direct gene counting. Conformity of the observed genotype frequencies with Hardy–Weinberg expectations (HWE) was examined by the exact test from Guo and Thompson [2] using the Arlequin software [3]. The parameters relevant for forensic casework (matching probability, power of exclusion, mean paternity index and

Table 1
Allele frequencies for 15 STR loci in a Flemish population sample ($n = 231$)

Allele	TH01	CSF1PO	D16S539	D13S317	D7S820	vWA	D8S1179	D21S11	D3S1358	D2S1338	D19S433	TPOX	D18S51	D5S818	FGA
5	0.002														
6	0.212														
7	0.154				0.011							0.002			
8	0.123		0.015	0.100	0.175		0.024					0.563		0.002	
9	0.139	0.030	0.115	0.080	0.165		0.009					0.084	0.002	0.037	
9.3	0.357														
10	0.013	0.260	0.069	0.043	0.264		0.104		0.002		0.002	0.045	0.004	0.030	
11		0.333	0.310	0.333	0.223		0.076				0.002	0.279	0.011	0.355	
12		0.305	0.301	0.279	0.134		0.126				0.067	0.026	0.136	0.377	
13		0.069	0.160	0.117	0.026		0.353		0.004		0.229		0.158	0.186	
13.2											0.013				
14		0.002	0.028	0.045	0.002	0.121	0.184		0.097		0.381		0.158	0.108	0.002
14.2											0.024				
15			0.002	0.002		0.095	0.106		0.223	0.002	0.182		0.145	0.002	
15.2											0.041				
16						0.234	0.017		0.262	0.035	0.037		0.117		
16.2											0.013				
17						0.229	0.002		0.212	0.208	0.006		0.115		
17.1									0.002						
17.2											0.002				
18						0.208			0.186	0.069			0.087		0.011
19						0.104			0.009	0.108			0.039		0.054
20						0.009			0.002	0.173			0.013		0.132
21										0.026			0.009		0.160
21.2															0.002
22										0.024			0.006		0.206
22.2															0.006
23										0.116					0.151
23.2															0.011
24										0.145					0.147
24.2															0.004
25										0.084					0.084
26								0.006		0.009					0.019
26.2															0.002
27								0.022							0.006
28								0.165							
29								0.208							
29.2								0.002							
30								0.249							
30.2								0.050							
31								0.078							
31.2								0.082							
32								0.035							
32.2								0.071							
33								0.002							
33.2								0.024							
34.2								0.006							

Table 2
Testing for HWE and statistical parameters of forensic interest (H_{obs}—observed heterozygosity, H_{exp}—expected heterozygosity, p value for exact test for HWE, MP—matching probability, PIC—polymorphism information content, PEX—power of exclusion, MPI—mean paternity index)

Stat.	STR locus														
	TH01	CSF1PO	vWA	D8 S1179	D21 S11	D7 S820	D3 S1358	D13 S317	D16 S539	D2 S1338	D19 S433	TPOX	D18 S51	D5 S818	FGA
H_{obs}	0.78	0.71	0.81	0.83	0.84	0.83	0.79	0.80	0.77	0.89	0.79	0.60	0.91	0.71	0.87
H_{exp}	0.77	0.72	0.82	0.80	0.85	0.81	0.79	0.78	0.77	0.87	0.76	0.60	0.88	0.70	0.86
p	0.78	0.70	0.93	0.16	0.50	0.02	0.84	0.95	0.54	0.33	0.20	0.10	0.61	0.88	0.56
MP	0.09	0.13	0.07	0.07	0.05	0.08	0.08	0.09	0.10	0.04	0.10	0.23	0.04	0.17	0.04
PIC	0.74	0.67	0.79	0.77	0.83	0.78	0.77	0.74	0.72	0.85	0.73	0.53	0.86	0.63	0.84
PEX	0.59	0.44	0.63	0.64	0.66	0.59	0.64	0.53	0.49	0.82	0.61	0.29	0.82	0.44	0.77
MPI	2.4	1.7	2.7	2.8	3.0	2.4	2.8	2.1	1.9	5.8	2.6	1.3	5.8	1.7	4.4

polymorphism information content) were determined using the Powerstat worksheet (Promega).

3. Results and discussion

Allelic frequencies in the Flemish population sample typed for the 15 Identifiler STRs are given in Table 1; results of testing for HWE and the statistical parameters of forensic interest are shown in Table 2. Regarding the test results for HWE, a p value >0.05 was obtained for all STRs except one. For D7S820 the exact test yielded a p value of 0.015. To judge whether to reject the null hypothesis (population equilibrium) based on the magnitude of the smallest of multiple p values, it is necessary to apply the Bonferroni [4] correction to the chosen significance threshold, which is typically 0.05. Considering the Bonferroni procedure and the fact that 15 tests for HWE were simultaneously performed on the same population sample, the significance threshold is adjusted from $\alpha = 0.05$ to $\alpha = 0.05/15 = 0.0033$ which is clearly below the p value of 0.015 that was observed for D7S820. Hence this single p value gives no reason to reject the null hypothesis.

Combined, the 15 STRs result in a matching probability of 1 in 111×10^{12} and a power of exclusion of 99.999995%, which should be effective in the resolution of most forensic and paternity cases.

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