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Allele frequency data for 15 STR loci (AmpFlSTR®, SGM plus™ and AmpFlSTR® Profiler™) in the Belgian population

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Abstract

The allele and genotype distributions for 15 STR loci included in the AmpFlSTR® SGM Plus™ and Profiler™ kits (Applied Biosystems) were determined in a sample of 100 Belgian individuals. No deviation from Hardy–Weinberg and genotypic equilibrium was observed. Four mutations were detected in 55 paternity and deficiency cases which indicated that the criteria for paternity exclusion should be adapted when a higher number of STR loci is used.

Keywords: STRs; Population database; Paternity testing; Mutation

1. Introduction

The use of multiplex PCR of STR loci with fluorescent detection of the alleles on automated capillary electrophoresis systems has significantly enhanced the ability to acquire genetic identity information in forensic DNA analysis and in paternity investigations.

In the past 10 years, our lab has gradually moved from a number of home-made VNTR/STR systems to the use of 13 STR loci (D3S1358, vWA, D16S539, D8S1179, D21S11, D18S51, THO1, FGA, TPOX, CSF1PO, D5S818, D13S317 and D7S820) in two multiplex PCR reactions (AmplFlSTR® Profiler Plus™ and COfiler™). Last year, we extended this set of STRs with two additional STRs (D2S1338 and D19S433) to further improve our ability to resolve complex kinship cases (e.g. immigration, deficiency cases). Here, we

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Table 1
Allele frequency and forensic efficiency data

Allele	D3S1358	VWA	D2S1338	D8S1179	D21S11	D18S51	D19S433	FGA
8				0.030				
9				0.010				
10				0.130		0.020		
11	0.005			0.070		0.020		
12				0.075		0.160	0.095	
13	0.005	0.005		0.285		0.160	0.210	
13.2							0.020	
14	0.135	0.080		0.265		0.155	0.375	
14.2							0.015	
15	0.245	0.120		0.105		0.140	0.150	
15.2							0.035	
16	0.210	0.205	0.050	0.030		0.120	0.060	
16.2							0.030	
17	0.210	0.290	0.165			0.060		
17.1	0.005							
17.2							0.010	
18	0.170	0.175	0.060			0.060		0.030
19	0.015	0.115	0.150			0.065		0.045
20		0.010	0.180			0.015		0.145
21			0.045			0.005		0.185
21.2								0.005
22			0.030			0.020		0.160
22.2								0.015
23			0.090					0.190
23.2								0.005
24			0.120					0.105
25			0.085					0.060
26			0.020		0.005			0.050
27					0.025			0.005
28			0.005		0.185			
29					0.240			
30					0.235			
30.2					0.025			
31					0.070			
31.2					0.075			
32					0.025			
32.2					0.080			
33					0.005			
33.2					0.030			
Heterozygosity	0.808	0.813	0.883	0.812	0.837	0.882	0.781	0.867
Matching probability	0.074	0.066	0.038	0.064	0.055	0.036	0.083	0.043
Power of exclusion	0.581	0.715	0.675	0.637	0.581	0.775	0.618	0.656

Table 1 (*continued*)

Allele	D16S539	THO1	TPOX	CSF1PO	D5S818	D13S317	D7S820
5		0.005					
6		0.225					
7		0.120					0.005
8	0.020	0.100	0.525			0.100	0.120
9	0.125	0.240	0.115	0.015	0.030	0.105	0.215
9.3		0.305					
10	0.050	0.005	0.040	0.255	0.045	0.055	0.290
11	0.325		0.280	0.320	0.300	0.290	0.200
12	0.300		0.040	0.310	0.410	0.265	0.125
13	0.170			0.075	0.185	0.115	0.045
14	0.010			0.020	0.030	0.065	
15				0.005		0.005	
Heterozygosity	0.761	0.778	0.633	0.734	0.707	0.808	0.802
Matching probability	0.102	0.088	0.219	0.139	0.143	0.065	0.080
Power of exclusion	0.493	0.398	0.413	0.581	0.383	0.637	0.715

report the results of a population study with the AmpFlSTR® SGM Plus™ and Profiler™ kits in a Belgian population.

2. Materials and methods

In total, 100 unrelated Caucasian individuals (34 females and 66 males) were selected from paternity cases. All individuals were known to be of Belgian descent and had been previously analysed with AmplFlSTR® Profiler Plus™ and COfiler™ kits (Applied Biosystems). DNA was extracted from blood by chelex extraction or from buccal swabs with the QiaAmp DNA Kit (Qiagen). The Amelogenin locus and 15 STRs were amplified with the AmpFlSTR® SGM Plus™ and the AmpFlSTR® Profiler™ kit (Applied Biosystems) using 25-μl reactions with 1 ng of DNA and cycling parameters according to the recommendations of the manufacturer. Amplified PCR products were size fractionated by means of capillary electrophoresis on the ABI PRISM® 310 Genetic Analyser (Applied Biosystems) with POP4 using GeneScan® 500[ROX] as internal size standard. Genotyping of the results was done with GeneScan® v2.1 and automated allele designation with Genotyper® v2.5 software (Applied Biosystems).

Allele and genotype frequencies, expected heterozygosity, Hardy–Weinberg equilibrium and genotypic disequilibrium were calculated with Genepop v3.1 software [1]. The matching probability and the power of exclusion were determined with Powerstats v1.2 (Promega) software.

3. Results and discussion

The number of alleles (Table 1) ranged from 5 (TPOX) to 13 (D18S51 and FGA) while expected heterozygosity levels were between 63.3% (TPOX) and 88.3% (D2S1338). No

Table 2
Observed STR mutations

STR	# Meiosis (paternal and maternal)[a]	Origin	
D2S1338[b]	76	Paternal	1-repeat loss (20 → 19)
D21S11	420	Maternal	1-repeat loss (33.2 → 32.2)
D7S820	203	Paternal[b]	1-repeat gain (11 → 12)
		Maternal	1-repeat gain or loss
		Maternal[b]	1-repeat gain or primer site mutation (10 → 11)[c]
CSF1PO[b]	203	Maternal	1-repeat gain or loss
vWA	420	Paternal	1-repeat loss (21 → 20)

[a] Accumulated data.
[b] This study.
[c] A silent allele cannot be excluded (mother and child both homozygous).

deviation from Hardy–Weinberg equilibrium was detected for all loci with an exact probability test. Exact tests for linkage disequilibrium between pairs of loci revealed no deviations even for the closely linked STRs CSF1PO and D5S818 which are localized, respectively on 5q33.3-34 and 5q23.3-32.

The combined power of exclusion for the 15 STRs was 0.9999992 while the combined matching probability was 1.7×10^{-17}. The AmpFLSTR® SGM Plus™ kit was more informative than the AmpFLSTR® Profiler™ kit which was mainly due to the presence of four highly polymorphic STR loci (D2S1338, D8S1179, D21S11 and D18S51).

The AmpFlSTR® SGM Plus™ and Profiler™ kits have been used so far in 52 paternity cases and 3 deficiency cases. Four mutations were observed (Table 2). In all cases, the mutation was the result of the loss or gain of one repeat unit. One maternal mutation observed for D7S820 could be due to the presence of a 'silent' allele or a mutation at one of the primer binding sites. Analysis with other primers than those of the AmpFlSTR® Profiler™ kit should enable us to distinguish between these different possibilities.

The observation of a 'high' number of mutations could be the result of the use of a high number (15) of STR loci. Therefore, the criteria for paternity exclusion should be adapted when the number of analysed loci is increased. Presently, we use at least three exclusions with at least one locus where all individuals tested are heterozygous.

Reference

[1] M. Raymond, F. Rousset, Genepop (version 1.2). A population genetic software for exact tests and ecumenicism, J. Hered. 86 (1995) 248–249.