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POPULATION DATA

Allelic proportions of 16 STR loci—including the new European Standard Set (ESS) loci—in a Swiss population sample

Christian Gehrig · Beate Balitzki · Adelgunde Kratzer · Christian Cossu · Naseem Malik · Vincent Castella

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Abstract Allele frequencies and forensically relevant population statistics of 16 STR loci, including the new European Standard Set (ESS) loci, were estimated from 668 unrelated individuals of Caucasian appearance living in different parts of Switzerland. The samples were amplified with a combination of the following three kits: AmpFlSTR® NGM SElect™, PowerPlex® ESI17 and PowerPlex® ESX 17. All loci were highly polymorphic and no significant departure from Hardy–Weinberg equilibrium and linkage equilibrium was detected after correction for sampling.

Keywords DNA · Multiplex STR · Forensic · Switzerland

Five Swiss laboratories, recognized by the Swiss Federal Department of Justice and Police for performing DNA analysis, have jointly decided to use a common local population database for statistical calculations. All five laboratories are accredited according to ISO/IEC 17025 and participate in national and international (ex. GEDNAP) proficiency tests. We determined the allelic proportions and forensic parameters for 16 STR autosomal loci, including the five new European Standard Set (ESS) loci [1], contained for instance in the AmpF*l*STR® NGM SElect™ (Applied Biosystems, USA) and the PowerPlex® ESI 17/ESX 17 (Promega, USA) kits. By publishing these data, our wish is to make them generally available.

Blood samples or buccal cell swabs were collected, after informed consent, from 668 unrelated individuals of Caucasian appearance living in different parts of Switzerland (Lausanne (n=200), Bern (n=202), Basel (n=21), Zurich (n=202) and St. Gallen (n=43)). The samples from Zurich and Lausanne were also used for European survey [2]. The different regions with the exception of Lausanne (French speaking) are all situated in the German-speaking part of the country. All regions are geographically very close (less than 200 km).

The laboratories have used different DNA extraction methods including iPrep, NaOH, Chelex and phenol–chloroform.

The samples from Basel were amplified only with the AmpF*l*STR® NGM SElect™ kit. The two laboratories from Bern and St. Gallen used a combination of the PowerPlex® ESI17 and PowerPlex® ESX 17 kits. The samples from Lausanne and Zurich were amplified using a combination of all the three aforementioned kits. Samples were amplified using 0.25–1.25 ng of target DNA using reaction conditions as described by each manufacturer (occasionally, a reduced PCR volume was used).

The amplified products were detected on the ABI PRISM 3100xl and 3130xl Genetic Analyzer (Applied Biosystems, USA) sequencers. Allele calling was performed using GeneMapper ID v3.2 (Applied Biosystems, USA) following the recommendations of the DNA Commission of the International Society of Forensic Genetics (IFSG) for the nomenclature of human STRs [3, 4].

C. Gehrig (✉) · V. Castella
Centre Universitaire Romand de Médecine Légale,
Geneva, Lausanne, Switzerland
e-mail: christian.gehrig@hcuge.ch

B. Balitzki
Institut für Rechtsmedizin Basel, Basel, Switzerland

A. Kratzer
Institut für Rechtsmedizin Zurich, Zurich, Switzerland

C. Cossu
Institut für Rechtsmedizin St. Gallen, St. Gallen, Switzerland

N. Malik
Institut für Rechtsmedizin Bern, Bern, Switzerland

Table 1 Allele frequencies for the NGM SElect/PowerPlex ESI 17 and PowerPlex ESX 17 kits in a Swiss population based on 668 individuals

Allele	D10S1248	vWA	D16S539	D2S1338	D8S1179	D21S11	D18S51	D22S1045	D19S443	TH01	FGA	D2S441	D3S1358	D1S1656	D12S391	SE33
5										0.001						
6										0.229						
6.2									0.001							
7										0.177						
8	0.001		0.019		0.012					0.124		0.001				
8.3										0.001						
9	0.001		0.141		0.008					0.166		0.001				0.001
9.3										0.289						
10			0.066		0.071		0.006		0.001	0.013		0.174		0.001		0.001
10.2							0.001									
11	0.004		0.313		0.095		0.016	0.144	0.004			0.339	0.001	0.070		
11.3												0.058				
12	0.026		0.281		0.127		0.146	0.009	0.077			0.051	0.001	0.130		0.002
12.1									0.001							
12.2									0.001							0.001
12.3												0.001				
13	0.294	0.001	0.162		0.303		0.142	0.005	0.250			0.025		0.077		0.016
13.2									0.016							0.003
14	0.312	0.100	0.017	0.001	0.223		0.174	0.046	0.341			0.290	0.111	0.093		0.026
14.2									0.019							0.001
14.3														0.004		
15	0.194	0.104	0.001	0.001	0.128		0.139	0.335	0.172			0.052	0.266	0.150	0.040	0.045
15.2									0.031							
15.3														0.062		
16	0.135	0.214		0.040	0.030		0.129	0.351	0.058			0.005	0.245	0.128	0.030	0.048
16.1																0.001
16.2									0.019		0.001					0.001
16.3														0.051		0.001
17	0.031	0.281		0.240	0.001		0.105	0.101	0.005				0.205	0.047	0.121	0.070
17.2									0.004							
17.3		0.026	0.001											0.116		
18	0.002	0.212		0.078	0.001		0.068	0.004			0.008		0.162	0.006	0.181	0.064
18.2									0.001							0.001
18.3														0.055	0.019	

Table 1 (continued)

Allele	D10S1248	vWA	D16S539	D2S1338	D8S1179	D21S11	D18S51	D22S1045	D19S443	TH01	FGA	D2S441	D3S1358	D1S1656	D12S391	SE33
19		0.075		0.115			0.039	0.003			0.059		0.008		0.108	0.076
19.2																0.002
19.3														0.008	0.013	
20		0.013		0.129			0.020	0.001			0.128		0.001		0.121	0.037
20.2																0.010
20.3														0.001	0.002	
21				0.038			0.010				0.189				0.115	0.030
21.2											0.002					0.010
21.3															0.001	
22				0.029			0.005				0.171				0.104	0.010
22.2											0.007					0.033
23				0.109			0.001				0.160				0.082	0.004
23.2											0.002					0.035
24				0.097							0.147				0.022	0.001
24.2											0.001					0.029
25				0.106							0.086				0.009	
25.2																0.033
26				0.016		0.003					0.031				0.004	
26.2																0.061
27				0.001		0.040					0.006				0.002	0.001
27.2																0.085
28						0.154					0.001					0.001
28.2																0.069
28.3																0.001
29						0.223										
29.2						0.001										0.078
30						0.251										
30.1																0.001
30.2						0.035										0.052
30.3																0.001
31						0.072										
31.2						0.097										0.028
32						0.014										0.001
32.2						0.081										0.014

Table 1 (continued)

Allele	D10S1248	vWA	D16S539	D2S1338	D8S1179	D21S11	D18S51	D22S1045	D19S443	TH01	FGA	D2S441	D3S1358	D1S1656	D12S391	SE33
33																0.002
33.2						0.022										0.002
34																0.005
34.2						0.004										
35																0.002
35.2						0.001										0.001
36																0.001
N	668	668	668	668	668	668	668	668	668	668	668	668	668	668	668	668
Obs. H	0.772	0.816	0.744	0.867	0.826	0.844	0.868	0.741	0.787	0.795	0.853	0.769	0.783	0.888	0.882	0.948
GD	0.759	0.804	0.772	0.871	0.811	0.839	0.875	0.732	0.781	0.790	0.860	0.761	0.789	0.900	0.892	0.948
PIC	0.719	0.776	0.737	0.857	0.786	0.820	0.861	0.688	0.750	0.757	0.844	0.725	0.755	0.891	0.882	0.945
PD	0.899	0.930	0.913	0.970	0.936	0.955	0.969	0.883	0.920	0.922	0.964	0.905	0.922	0.980	0.977	0.993
PE	0.549	0.629	0.500	0.728	0.649	0.684	0.731	0.495	0.576	0.590	0.701	0.544	0.568	0.770	0.758	0.893

Obs. H observed heterozygosity, *GD* gene diversity, *PIC* polymorphism information content, *PD* power of discrimination, *PE* power of exclusion

For concordance studies, 245 samples (Bern and St. Gallen) were typed each with PowerPlex® ESI 17 and PowerPlex® ESX 17 and another 402 (Lausanne and Zurich) were typed using the same three kits (PowerPlex® ESI 17, PowerPlex® ESX 17 and AmpF*l*STR® NGM SElect™). A comparison of all results revealed no differences between the genotypes obtained using the different kits.

Allelic proportions and forensic parameters were calculated using PowerStats version 1.2 [5] (see Table 1). Fst values, gene diversity and *P* values for Hardy–Weinberg equilibrium were assessed using FSTAT (http://www.unil.ch/Jahia/site/dee/op/edit/pid/36921).

The *P* value for Hardy–Weinberg proportions, when using the Bonferroni correction, was assigned as $P<0.05/15=0.0033$. As suggested by Buckleton et al. [6], we do not comment on the value itself, indeed 'It is not likely that Hardy-Weinberg disequilibrium, at the level it is thought to exist in human populations, will be detected with samples of 1000 or less'.

The observed heterozygosity varies between 0.741 for D22S1045 and 0.948 for SE33. The gene diversity (also termed Hex) was smallest for D22S1045 (0.732) and largest for SE33 (0.948). The power of discrimination ranges from 0.883 for D22S1045 to 0.993 for SE33. Before pooling the data, we calculated coancestry values (Fst) values for the 16 NGM SElect loci for the five Swiss population groups reported here (Table 2). The maximum Fst was observed at the D3S1358 locus (i.e. 0.003). Altogether, based on these data and on the general knowledge of the population substructure in Europe, we decided

Table 2 Fst values based on five Swiss population samples

	Fst
D3S1358	0.003
D19S433	−0.001
D2S1338	−0.001
D22S1045	0.000
D16S533	0.001
D18S51	0.000
D1S1656	0.000
D10S1248	0.001
D2S441	−0.001
TH01	−0.001
vWA	−0.000
D21S11	−0.001
D12S391	−0.001
D8S1179	−0.002
FGA	0.001
SE33	0.000
Avg	−0.000

to group the allelic frequency data of the different Swiss regions together in order to obtain national population data.

Our samples from Zurich ($n=202$) and Lausanne ($n=200$) were previously compared to 24 European populations [2]. This study showed that the genetic variability is evenly distributed among these populations. Regarding the value of theta itself, we recommend to use the widely accepted value of 0.01.

In conclusion, we believe that for cases where the relevant population is Switzerland, these data will be useful; thus, our wish is to make them available publicly. This paper follows the guidelines for publication of population data requested by the journal [7].

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