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Short Communication

Population genetic study for 24 STR loci and Y indel (GlobalFiler™ PCR Amplification kit and PowerPlex® Fusion system) in 1000 Korean individuals

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ABSTRACT

Allele frequencies for 23 autosomal short tandem repeat loci (D3S1358, vWA, D16S539, CSF1PO, TPOX, D8S1179, D21S11, D18S51, TH01, FGA, D5S818, D13S317, D7S820, D2S441, D19S433, D22S1045, D10S1248, D1S1656, D12S391, D2S1338, SE33, Penta D, Penta E), 1 Y-chromosome short tandem repeat locus (DYS391) and Y indel were obtained from 1000 unrelated individuals of the Korean population.

1. Population

In this study, we analyzed a total of 1000 unrelated individuals (497 males and 503 females) to calculate the population allele frequency. DNA samples were obtained from the Korea Biobank in Korea Centers for Disease Control & Prevention, Republic of Korea. The use of the samples and the analytical procedures were approved by the Ethics Committee and Institutional Review Board of Yonsei University in Korea.

2. PCR

For PCR amplification, we followed the manufacturers' recommended protocols for GlobalFiler™ PCR Amplification kit (Applied Biosystems, Foster City, CA, USA) and PowerPlex® Fusion system (Promega, Madison, WI, USA).

3. Genotyping

GeneScan 600 LIZ size standard (Life Technologies) and CC5 Internal lane Standard 500 (Promega) were used for capillary electrophoresis of GlobalFiler™ PCR Amplification kit and PowerPlex® Fusion system, respectively. The electrophoresis was run on a 3500xl genetic analyzer (Life Technologies). Allele designations were named according to the recommendations of the DNA Commission of the International Society of Forensic Genetics (ISFG) [1,2].

4. Analysis of data

The data were analyzed using the GeneMapper ID-X software v1.2 (Life Technologies) and Microsoft Excel (Microsoft, Redmond, WA, USA). The exact test was performed for assessing the Hardy-Weinberg equilibrium (HWE) using Arlequin v3.5 as dememorization steps of 10,000 iterations per batch and 1,000,000 steps in Markov chain [3,4]. And 1000 batches were performed for HWE. Statistical analysis, including allele frequencies, heterozygosities, polymorphism information content (PIC), and genetic distances, were performed with PowerMarker v3.25 software [5]. Genotyping results were compared for the concordance test to determine if there were any discordant allele calls between the GlobalFiler™

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PCR Amplification kit and PowerPlex® Fusion system. Important parameters for forensic paternity testing were calculated using PowerStats v1.2 [6]. Linkage disequilibrium tests between pairs of loci on the same chromosome were performed using Arlequin v3.5 based on a likelihood ratio test for unknown gametic phase, whose empirical distribution is obtained from 10,000 permutations [4].

5. Results

See Tables 1–4.

5.1. Other remarks

The exact test showed no significant deviations from HWE, except at the FGA locus ($p = 0.026$). However, FGA was not significant after Bonferroni correction for multiple testing ($p < 0.05/23 = 0.002174$). The most informative locus was SE33 ($H_{obs} = 0.948$), whereas TPOX was the least informative locus ($H_{obs} = 0.626$).

The combined PM calculated from the 21 autosomal STR loci included in the GlobalFiler™ PCR Amplification kit was 2.937×10^{-25}, and that from the 22 autosomal STR loci included in the PowerPlex® Fusion system was 3.601×10^{-26}. This value was higher than the combined PM of 22 loci analyzed with the PowerPlex® Fusion system in the Japanese population (2.93×10^{-26}) [7]. The combined PM from the 24 STR loci for both kits was 2.902×10^{-28}, and the combined PE from the 24 loci was 0.9999999971, which could be considered highly informative. Although SE33 was the most informative (PD = 0.992) and was included only in the GlobalFiler™ PCR Amplification kit, the discriminating power was higher in the PowerPlex® Fusion system since the PowerPlex® Fusion system had one more locus than the GlobalFiler™ PCR Amplification kit.

The 23 autosomal STR loci included in the GlobalFiler™ PCR Amplification kit and PowerPlex® Fusion system were located on seventeen chromosomes. D12S391 and vWA both resided on chromosome 12; D2S441, TPOX, and D2S1338 were on chromosome 2; D5S818 and CSF1PO were on chromosome 5; and D21S11 and Penta D were found on chromosome 21. Thus, we performed linkage disequilibrium tests for loci on the same chromosome. No linkage disequilibrium was observed among each pair of loci ($p > 0.05$, Table 3).

Concordance studies are necessary to access the frequency of silent allele or one or more nucleotides insertion/deletion in a dataset when different primer sets are used for the same samples [8,9]. These errors would give rise to incorrect exclusion of two samples from the same source. Thus, we compared a total of 20,000 genotypes (20 loci × 1000 samples) between the GlobalFiler™ PCR Amplification kit and PowerPlex® Fusion system for concordance evaluation (Table 4). The concordant rate was 99.975% (19,995 out of 20,000 genotypes). We observed four null alleles due to silent allele at the D19S433 locus for the GlobalFiler™ PCR Amplification kit in three samples and at the TPOX for the

Table 1
Forensic parameters for the 23 autosomal STR loci other than DYS391 and Y indel in 1000 Korean individuals.

	D3S1358	vWA	D16S539	CSF1PO	TPOX	D8S1179
N	1000	1000	1000	1000	1000	1000
H_{obs}	0.699	0.796	0.779	0.686	0.626	0.832
H_{exp}	0.704	0.797	0.778	0.724	0.624	0.844
Exact test (*p*)	0.399	0.605	0.332	0.075	0.147	0.319
PM	0.137	0.074	0.087	0.117	0.216	0.044
PD	0.863	0.926	0.913	0.883	0.784	0.956
PIC	0.655	0.766	0.743	0.679	0.556	0.825
PE	0.427	0.592	0.561	0.407	0.323	0.660
	D21S11	D18S51	D2S441	D19S433	TH01	FGA
N	1000	1000	1000	1000	1000	1000
H_{obs}	0.796	0.849	0.785	0.786	0.668	0.826
H_{exp}	0.791	0.859	0.776	0.795	0.671	0.848
Exact test (*p*)	0.438	0.972	0.581	0.250	0.066	0.026
PM	0.070	0.035	0.083	0.069	0.158	0.041
PD	0.930	0.965	0.917	0.931	0.842	0.959
PIC	0.766	0.844	0.745	0.768	0.622	0.831
PE	0.592	0.693	0.572	0.573	0.381	0.648
	D22S1045	D5S818	D13S317	D7S820	D10S1248	D1S1656
N	1000	1000	1000	1000	1000	1000
H_{obs}	0.745	0.772	0.780	0.754	0.740	0.820
H_{exp}	0.759	0.774	0.805	0.768	0.754	0.828
Exact test (*p*)	0.842	0.435	0.230	0.147	0.798	0.605
PM	0.099	0.085	0.065	0.088	0.100	0.049
PD	0.901	0.915	0.935	0.912	0.900	0.951
PIC	0.717	0.740	0.777	0.734	0.715	0.808
PE	0.501	0.548	0.562	0.517	0.493	0.637
	D12S391	D2S1338	SE33	Penta D	Penta E	
N	1000	1000	1000	1000	1000	
H_{obs}	0.831	0.870	0.948	0.789	0.940	
H_{exp}	0.822	0.871	0.940	0.791	0.919	
Exact test (*p*)	1.000	0.389	0.132	0.594	0.083	
PM	0.055	0.031	0.008	0.073	0.014	
PD	0.945	0.969	0.992	0.927	0.986	
PIC	0.799	0.857	0.936	0.763	0.914	
PE	0.658	0.735	0.894	0.579	0.878	

H_{obs}, observed heterozygosity; H_{exp}, expected heterozygosity; PM, probability of a match; PD, power of discrimination; PIC, polymorphism information content; PE, paternity power of exclusion.