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Population genetics data for 22 autosomal STR loci in European, South Asian and African populations using SureID® 23comp Human DNA Identification Kit

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Abstract

Allele frequency data for 22 short tandem repeat loci; D18S1364, D1S1656, D13S325, D5S2800, D9S1122, D4S2366, D3S1744, D12S391, D11S2368, D21S2055, D20S482, D8S1132, D7S3048, D2S441, D19S253, D10S1248, D17S1301, D22-GATA198B05, D16S539, D6S474, D14S1434 and D15S659 from the SureID® 23comp Human DNA Identification Kit have been determined for unrelated individuals in European, South Asian and African populations. Deviations from Hardy–Weinberg equilibrium were observed in loci D1S1656 and D19S253 in European; D18S1364, D6S474 and D14S1434 in South Asian; and D9S1122 and D8S1132 in African populations (p -value < 0.05). However, after Bonferroni correction no significant deviations were observed (p -value < 0.002). The most discriminating loci were D1S1656 and D12S391 for European ($PD = 0.977$), D21S2055 for South Asian ($PD = 0.980$), and D21S2055 and D7S3048 for African ($PD = 0.972$) populations. The match probabilities were 1 in 6.7×10^{25} for European, 1 in 1.4×10^{26} for South Asian and 1 in 1.6×10^{26} for African populations. These findings established the high discriminatory capacity and robustness of the tested STR loci for forensic identification and kinship testing.

Introduction

Over the years, several STR multiplex kits which comprise different complementary molecular markers have been optimised and used for human identification and kinship testing. Complementary kits that comprise STR loci outside of the European Standard Set (ESS) [1] and Combined DNA Index System (CODIS) [2] STR loci standards provide value in complex case analysis where routine kits do not generate enough data for a conclusive report. Such cases can include but are not limited to; close kinship, incest and mixed samples. Amongst other uses, forensic applications of these kits require the generation of allele frequency databases specific for the populations where they would be used [3–5], rather than use of global database which might not be a true representation of an individual.

Currently, Anglia DNA Services uses the AmpF ℓ STR[®] Identifiler[®] PCR Amplification kit (Applied Biosystems[®]) for parentage and other relationship testing [6–8]. To improve the customer charter by reducing the turn-around time and the number of inconclusive results, a robust and advanced multiplex; SureID[®] 23comp Human DNA Identification Kit (Ningbo Health Gene Technologies) was tested in our laboratory. This kit comprises 22 autosomal STR markers compared to 15 present in the AmpF ℓ STR[®] Identifiler[®] kit. Both kits also comprise a sex-determining marker. Ten loci in the SureID[®] 23comp kit; D1S1656, D4S2366, D3S1744, D12S391, D21S2055, D8S1132, D2S441, D10S1248, D16S539, and D6S474 were observed to overlap in few other popular kits such as GlobalFiler[™] PCR Amplification Kit (Applied Biosystems[®]), PowerPlex[®] Fusion System (Promega) and Investigator[®] HDplex kit (Qiagen[®]). Currently, allele frequencies for D18S1364, D13S325, D5S2800, D11S2368, D7S3048, D19S253, D22-GATA198B05 and D15S659 loci for European, South Asian and African populations are not publicly available on the popular population databases; the European Network of Forensic Science Institutes (ENFSI) DNA working group STR population database (STRidER; <http://strider.online>) [9], the National Institute of Standards and Technology (NIST) Population Dataset (STRBase; <https://strbase.nist.gov/NISTpop.htm>) [10], ALFRED, the ALlele FREquency Database (<https://alfred.med.yale.edu/>) [11], *pop.STR* (<http://spsmart.cesga.es/popstr.php>) [12], PopAffiliator (<http://cracs.fc.up.pt/~nf/popaffiliator2>) [13] and ALLST*R (<http://allstr.de>). The aim of this study was to develop allele frequency databases for these three populations which would be useful for human identification and forensic casework applications.

Materials and methods

Population samples

Buccal swabs of 920 anonymous unrelated individuals (321 Europeans, 315 South Asians and 284 Africans) with self-proclaimed ethnicities for three generations were selected from the casework samples for this validation study. Informed consent was obtained during sampling under Anglia DNA Services' terms and conditions [14].

DNA extraction and PCR amplification

DNA from buccal swabs was extracted using the QIAamp[®] DNA Minikit (Qiagen[®]) and amplified according to the manufacturer's recommendations for the SureID[®] 23comp kit. Amplification was carried out using GeneAmp[®] PCR System 9700 thermal cycler (Applied Biosystems[®]).

Electrophoresis and typing

Capillary electrophoresis was performed using the ABI Prism® 3500xL Genetic Analyzer (Applied Biosystems®) and alleles were determined using GeneMapper® ID-X v1.3 software (Applied Biosystems®).

Quality controls

Control DNA and allelic ladders provided by the manufacturer of the SureID® 23comp kit were used for quality assurance. Thirty random samples from the population samples (28 Europeans and 2 Africans) were also amplified with Investigator® HDplex kit (Qiagen®) for concordance study between common loci (D12S391, D3S1744, D4S2366, D6S474, D8S1132 and D21S2055).

Analysis of data

Forensic and population parameters of the 22 STRs were estimated with PowerStats ver. 1.2 (Promega) and the p-values of exact tests for Hardy–Weinberg equilibrium and population differentiation were generated using Arlequin ver. 3.5 [15].

Results and discussion

Population allele frequencies and forensic parameters are presented in Tables 1–6. Loci D1S1656 (p-value = 0.026) and D19S253 (p-value = 0.015) in European; D18S1364 (p-value = 0.012), D6S474 (p-value = 0.017) and D14S1434 (p-value = 0.043) in South Asian; and D9S1122 (p-value = 0.006) and D8S1132 (p-value = 0.047) in African populations showed significant deviations from Hardy–Weinberg equilibrium (p-value < 0.05). However, no significant deviations were observed after Bonferroni correction (p-value < 0.002). These deviations could be due to population size, inbreeding, population substructure or selection. Another important factor could be the self-declaration of ethnicity by the sample donors which could be biased. This has been previously reported in the study of Walsh et al. [16], where the authors assessed the effect of self-declaration on the data generated and its fitness of purpose for forensic applications. The authors established that the expected genetic pattern was observed and although not perfect, the database developed was applicable for forensic use.

In this study, the most discriminating loci were D1S1656 and D12S391 for European (PD = 0.977), D21S2055 for South Asian (PD = 0.980), and D21S2055 and D7S3048 for African (PD = 0.972) populations. The match probabilities were 1 in 6.7×10^{25} for European, 1 in 1.4×10^{26} for South Asian and 1 in 1.6×10^{26} for African populations. The inter-population comparisons revealed no significant differences between all three populations (p-value < 0.002).

The comparison study on the common loci with Investigator® HDplex kit showed concordant results except for locus D6S474 where the called alleles were 1-repeat less (e.g. 13, 15 on Investigator®

HDplex but 14, 16 on SureID® 23comp). Discordance between different STR kits have been reported previously [17–20], thus it is recommended that careful approach should be taken when comparing results or using population databases generated using other kits for SureID® 23comp kit profiles.

Conclusion

A total of three different populations have been studied with their corresponding allele frequencies generated. Overall, results obtained establish high discriminatory capacity and robustness of the tested STR loci for forensic identification and kinship testing for the European, South Asian and African populations.

This paper follows the guidelines for publication of population data as described by the journal [21–23].

Declaration of interest

The authors declared no conflicts of interest.

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Allele	D18S1364	D1S1656	D13S325	D5S2800	D9S1122	D4S2366	D3S1744	D12S391	D11S2368	D21S2055	D20S482	D8S1132	D7S3048	D2S441	D19S253	D10S1248	D17S1301	D22- GATA198B05	D16S539	D6S474	D14S1434	D15S659
7														0.002	0.268							
8		0.002												0.002	0.034		0.002		0.008			
9					0.002	0.347					0.011			0.002	0.014		0.002		0.125			0.005
10		0.002			0.022	0.178								0.174	0.020		0.019		0.070		0.192	0.020
11	0.002	0.081			0.192	0.092					0.016			0.371	0.086	0.003	0.346		0.322		0.041	0.192
11.3														0.042								
12	0.006	0.137			0.399	0.157					0.039			0.036	0.343	0.039	0.417		0.279		0.019	0.217
12.1												0.002										
13	0.201	0.070			0.330	0.126					0.229			0.039	0.160	0.312	0.185		0.167	0.003	0.357	0.058
13.1														0.002								
13.3														0.002								
14	0.246	0.084		0.291	0.047	0.092	0.106				0.430			0.274	0.062	0.301	0.028		0.025	0.232	0.374	0.031
14.3		0.008	0.003																			
15	0.301	0.151			0.008	0.008	0.084	0.041	0.002		0.209	0.002	0.002	0.048	0.012	0.187	0.002	0.003	0.005	0.196	0.019	0.157
15.3		0.070																				
16	0.103	0.081	0.022		0.002		0.125	0.025	0.003		0.065	0.012	0.005	0.008		0.128		0.117		0.151		0.190
16.1										0.053												
16.3		0.048																				
17	0.026	0.042	0.022	0.344			0.296	0.129	0.039		0.002	0.132	0.028			0.028		0.134		0.282		0.107
17.1										0.014												
17.3		0.148						0.020														
18	0.079	0.009	0.053	0.246			0.206	0.173	0.148			0.196	0.059			0.003		0.131		0.123		0.022
18.1										0.008												
18.3		0.064						0.036														
19	0.028		0.235	0.003			0.122	0.123	0.240			0.146	0.073					0.192		0.011		0.002
19.1										0.259												
19.3		0.003						0.012														
20	0.006		0.248	0.009			0.047	0.112	0.245			0.154	0.092					0.143		0.002		
20.1										0.042												
20.3			0.005																			
21	0.002		0.182				0.014	0.101	0.210	0.003		0.122	0.153					0.201				
21.1										0.005												
22			0.146	0.003			0.002	0.101	0.076			0.126	0.120					0.072				
22.2												0.002										
23			0.069	0.086				0.073	0.026	0.006		0.076	0.179					0.006				
24			0.014	0.017				0.036	0.006	0.016		0.023	0.145					0.002				
25			0.002					0.016	0.005	0.125		0.006	0.114									
26								0.002		0.162			0.025									
27										0.006			0.006									
28										0.020												
29										0.030												
30										0.016												
31										0.028												
32										0.025												
33										0.058												
34										0.069												
35										0.044												
36										0.012												
37										0.002												

Table 2: Hardy–Weinberg equilibrium evaluation and forensic parameters for 22 STR loci in the European population.

Locus	D18S1364	D1S1656	D13S325	D5S2800	D9S1122	D4S2366	D3S1744	D12S391	D11S2368	D21S2055	D20S482	D8S1132	D7S3048	D2S441	D19S253	D10S1248	D17S1301	D22- GATA198B05	D16S539	D6S474	D14S1434	D15S659
Ho	0.782	0.885	0.854	0.710	0.685	0.776	0.829	0.879	0.813	0.838	0.688	0.872	0.882	0.776	0.710	0.785	0.636	0.829	0.769	0.788	0.679	0.819
He	0.791	0.900	0.821	0.729	0.694	0.791	0.820	0.896	0.810	0.875	0.714	0.863	0.879	0.751	0.773	0.760	0.672	0.850	0.771	0.791	0.695	0.840
P	0.954	0.026	0.551	0.695	0.297	0.739	0.999	0.431	0.862	0.102	0.464	0.845	0.152	0.753	0.015	0.877	0.646	0.196	0.592	0.111	0.957	0.543
PD	0.927	0.977	0.940	0.881	0.848	0.927	0.945	0.977	0.934	0.971	0.876	0.962	0.968	0.898	0.919	0.897	0.836	0.956	0.907	0.918	0.855	0.953
PE	0.566	0.764	0.702	0.444	0.406	0.555	0.653	0.752	0.624	0.671	0.411	0.739	0.758	0.555	0.444	0.572	0.336	0.653	0.544	0.577	0.397	0.635
MP	0.073	0.023	0.060	0.119	0.152	0.073	0.055	0.023	0.066	0.029	0.124	0.038	0.032	0.102	0.081	0.103	0.164	0.044	0.093	0.082	0.145	0.047
PIC	0.760	0.889	0.796	0.680	0.637	0.763	0.797	0.885	0.781	0.863	0.669	0.846	0.865	0.713	0.740	0.720	0.608	0.830	0.734	0.757	0.637	0.818
TPI	2.293	4.338	3.415	1.726	1.589	2.229	2.918	4.115	2.675	3.087	1.605	3.915	4.224	2.229	1.726	2.326	1.372	2.918	2.169	2.360	1.558	2.767

Ho: observed heterozygosity, He: expected heterozygosity, P: Hardy–Weinberg exact test, PD: power of discrimination, PE: power of exclusion, PIC: polymorphism information content, TPI: typical paternity index.

Table 3: Allele frequencies for 22 STR loci in the South Asian population (n = 315).

Allele	D18S1364	D1S1656	D13S325	D5S2800	D9S1122	D4S2366	D3S1744	D12S391	D11S2368	D21S2055	D20S482	D8S1132	D7S3048	D2S441	D19S253	D10S1248	D17S1301	D22- GATA198B05	D16S539	D6S474	D14S1434	D15S659
6															0.002							
7															0.248							0.005
8		0.032													0.035				0.057			0.005
9		0.006				0.284					0.003				0.008		0.005		0.152			0.014
10		0.014			0.025	0.163					0.005			0.273	0.030		0.013		0.119		0.133	0.052
10.1												0.002										
11		0.152			0.249	0.160	0.002				0.008			0.433	0.124	0.016	0.216		0.325		0.092	0.200
11.3														0.049								
12	0.017	0.084			0.378	0.156	0.003				0.044			0.060	0.308	0.017	0.535		0.216		0.052	0.144
12.1												0.005										
12.3														0.006								
13	0.263	0.119		0.002	0.302	0.129					0.224			0.011	0.176	0.167	0.194		0.119	0.005	0.356	0.073
13.1												0.003										
14	0.165	0.086		0.279	0.037	0.098	0.084		0.002		0.478			0.154	0.060	0.306	0.035		0.011	0.254	0.343	0.032
14.1												0.002										
14.3			0.002																	0.003		
15	0.235	0.184		0.003	0.006	0.010	0.071	0.006			0.186			0.011	0.010	0.241	0.003	0.008		0.321	0.017	0.179
15.3		0.024																				
16	0.110	0.141	0.008	0.010	0.003		0.140	0.016	0.016		0.051	0.005	0.003	0.002		0.186		0.132		0.133	0.003	0.170
16.1										0.146												
16.3		0.021																				
17	0.048	0.060	0.005	0.262			0.271	0.137	0.049			0.076	0.044			0.062		0.229		0.173	0.002	0.097
17.1										0.013												
17.3		0.043						0.003														
18	0.113	0.016	0.056	0.292			0.192	0.238	0.121		0.002	0.216	0.044			0.005		0.078		0.100	0.002	0.024
18.1										0.025												
18.3		0.013						0.016														
19	0.038	0.002	0.237	0.010			0.173	0.165	0.216			0.197	0.094					0.124		0.011		0.005
19.1										0.110												
19.3		0.003																				
20	0.010		0.251	0.044			0.057	0.103	0.243			0.154	0.159					0.156				
20.1										0.011												
21	0.002		0.203				0.005	0.097	0.233			0.125	0.137					0.190				
21.1										0.006												
22			0.148	0.002				0.114	0.086			0.097	0.098					0.073				
23			0.065	0.067			0.002	0.071	0.029	0.002		0.086	0.184					0.010				
24			0.019	0.030				0.025	0.005	0.014		0.030	0.113					0.002				
24.3			0.002																			
25			0.005					0.006		0.175		0.003	0.090									
26								0.002	0.002	0.140			0.030									
26.3			0.002																			
27										0.017			0.003									
28										0.017												
29										0.027												
30										0.025												
31										0.040												
32										0.052												
33										0.048												
34										0.076												
35										0.044												
36										0.005												
37										0.006												

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Table 4: Hardy–Weinberg equilibrium evaluation and forensic parameters for 22 STR loci in the South Asian population.

Locus	D18S1364	D1S1656	D13S325	D5S2800	D9S1122	D4S2366	D3S1744	D12S391	D11S2368	D21S2055	D20S482	D8S1132	D7S3048	D2S441	D19S253	D10S1248	D17S1301	D22- GATA198B05	D16S539	D6S474	D14S1434	D15S659
Ho	0.756	0.905	0.797	0.708	0.695	0.800	0.803	0.825	0.822	0.863	0.679	0.800	0.876	0.679	0.778	0.781	0.629	0.832	0.775	0.717	0.708	0.825
He	0.821	0.888	0.812	0.762	0.703	0.818	0.826	0.859	0.816	0.901	0.684	0.853	0.879	0.709	0.793	0.783	0.629	0.844	0.794	0.776	0.728	0.860
P	0.012	0.248	0.467	0.165	0.940	0.465	0.206	0.197	0.149	0.589	0.684	0.133	0.681	0.392	0.195	0.106	0.065	0.480	0.285	0.017	0.043	0.460
PD	0.943	0.972	0.936	0.910	0.856	0.941	0.945	0.963	0.935	0.980	0.855	0.960	0.971	0.873	0.925	0.916	0.807	0.955	0.926	0.915	0.875	0.963
PE	0.519	0.805	0.593	0.441	0.421	0.599	0.605	0.647	0.641	0.722	0.397	0.599	0.747	0.397	0.558	0.564	0.327	0.659	0.553	0.456	0.441	0.647
MP	0.057	0.028	0.064	0.090	0.144	0.059	0.055	0.037	0.065	0.020	0.145	0.040	0.029	0.127	0.075	0.084	0.193	0.045	0.074	0.085	0.125	0.037
PIC	0.796	0.876	0.784	0.721	0.646	0.791	0.802	0.843	0.789	0.891	0.638	0.834	0.866	0.664	0.762	0.748	0.576	0.824	0.764	0.740	0.682	0.843
TPI	2.045	5.250	2.461	1.712	1.641	2.500	2.540	2.864	2.813	3.663	1.559	2.500	4.038	1.559	2.250	2.283	1.346	2.972	2.218	1.770	1.712	2.864

Ho: observed heterozygosity, He: expected heterozygosity, P: Hardy–Weinberg exact test, PD: power of discrimination, PE: power of exclusion, PIC: polymorphism information content, TPI: typical paternity index.

Table 5: Allele frequencies for 22 STR loci in the African population (n = 284).

[illegible]

[illegible]

Table 6: Hardy–Weinberg equilibrium evaluation and forensic parameters for 22 STR loci in the African population.

Locus	D18S1364	D15S1656	D13S325	D5S2800	D9S1122	D4S2366	D3S1744	D12S391	D11S2368	D21S2055	D20S482	D8S1132	D7S3048	D2S441	D19S253	D10S1248	D17S1301	D22- GATA198B05	D16S539	D6S474	D14S1434	D15S659
Ho	0.880	0.877	0.813	0.764	0.722	0.768	0.729	0.835	0.799	0.870	0.690	0.870	0.873	0.775	0.849	0.775	0.651	0.859	0.771	0.743	0.687	0.778
He	0.869	0.850	0.809	0.785	0.716	0.749	0.773	0.868	0.822	0.883	0.698	0.882	0.883	0.769	0.834	0.796	0.633	0.846	0.802	0.775	0.722	0.837
P	0.682	0.452	0.550	0.752	0.006	0.430	0.399	0.225	0.522	0.230	0.751	0.047	0.838	0.461	0.747	0.543	0.988	0.414	0.510	0.379	0.452	0.221
PD	0.965	0.957	0.931	0.920	0.862	0.889	0.920	0.967	0.942	0.972	0.875	0.970	0.972	0.908	0.946	0.929	0.808	0.953	0.931	0.912	0.875	0.956
PE	0.755	0.748	0.624	0.534	0.463	0.540	0.474	0.665	0.598	0.734	0.413	0.734	0.741	0.553	0.692	0.553	0.357	0.713	0.547	0.498	0.408	0.559
MP	0.035	0.043	0.069	0.080	0.138	0.111	0.080	0.033	0.058	0.028	0.125	0.030	0.028	0.092	0.054	0.071	0.192	0.047	0.069	0.088	0.125	0.044
PIC	0.853	0.833	0.780	0.750	0.664	0.708	0.745	0.853	0.796	0.871	0.662	0.869	0.869	0.734	0.812	0.765	0.577	0.825	0.772	0.737	0.673	0.817
TPI	4.176	4.057	2.679	2.119	1.797	2.152	1.844	3.021	2.491	3.838	1.614	3.838	3.944	2.219	3.302	2.219	1.434	3.550	2.185	1.945	1.596	2.254

Ho: observed heterozygosity, He: expected heterozygosity, P: Hardy–Weinberg exact test, PD: power of discrimination, PE: power of exclusion, PIC: polymorphism information content, TPI: typical paternity index.