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FOR THE RECORD

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Genetic Analysis of Large Data Sets of North American Black, Caucasian, and Hispanic Populations at 13 CODIS STR Loci

POPULATIONS: United States, Black, Caucasian, Hispanic

KEYWORDS: forensic science, United States, Black, Caucasian, Hispanic, population genetics, allele frequency, DNA typing, D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D7S820, D16S539, TH01, TPOX and CSF1PO

Over 300,000 blood or buccal specimens were obtained from North American individuals in order to generate genetic data to help determine biological relationships. Genomic DNA was isolated from each specimen using phenol-chloroform extraction or other standard methodologies (1) and quantitated using PICO green (Molecular Probes). Approximately 1 ng of each DNA sample was subjected to PCR amplification with both the Applied Biosystems (ABI) AmpFlSTR Profiler Plus and Cofiler systems. These test batteries provide genotype information for the 13 distinct Combined DNA Index System (CODIS) short tandem repeat (STR) loci (D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D7S820, D16S539, TH01, TPOX and CSF1PO) used widely in forensic and paternity applications. The fluorescently-labeled amplification products were analyzed by denaturing polyacrylamide gel electrophoresis on an ABI Prism 377 DNA sequencer. The gels were analyzed by GeneScanner software and allele calls were made using the Genotyper program (both from ABI).

The resultant genetic data were electronically archived and three separate database queries were performed: one for 8000 genotypes (16,000 alleles/locus) of Blacks, another for 8000 genotypes of Caucasian individuals and the last for 1000 genotypes of Hispanics (2000 alleles/locus). Relatively fewer genotypes of Hispanics were included in this analysis due in part to the under-representation of Hispanic individuals submitting to genetic testing for cases of disputed paternity compared to Caucasians and Blacks. Furthermore, only Hispanics genotyped prior to calendar year 2001 were included in the database output because many Hispanic individuals typed after that date originated from a population potentially comprised of significant substructure. Additionally, a search protocol was designed for each of the three databases such that only those individuals entered as either a mother or an alleged father involved in a disputed paternity case would be included in the output file because such individuals are not likely biologically related. Finally, it was stipulated that a given person would only be included once in each output file regardless of whether multiple specimens from that individual were tested and the data uploaded, thus representing more than a single iteration of that particular genotype within the database.

Output data from each population were analyzed by Genetic Data Analysis (GDA) software (2). Allele frequencies were calculated at each locus by counting the numbers of each genotype present in the sample set. Unbiased estimates of expected heterozygosity frequencies were also calculated along with observed departures from Hardy-Weinberg equilibrium expectations as computed by the Fisher exact test based on 2000 shufflings (3). Probability of exclusion and power of discrimination statistics are also provided for each STR locus within each population.

The observed allele frequencies for each locus in each population are shown in Tables 1–3. FGA and D18S51 are the most informative systems in each of the 3 populations. The least discriminating loci are D13S317 and TH01 in North American Blacks and D5S818 and TPOX in Caucasians and Hispanics. TPOX ($p = 0.042$) in Caucasians and vWA ($p = 0.019$) in Blacks are the only loci to depart from Hardy-Weinberg expectations based on the exact test but do not comprise significant departures after correction for sampling (4). The combined powers of discrimination of the 13 CODIS loci exceed 0.9999999999999 in each of the three populations.

The complete data set can be accessed by any interested party at http://www/technology/publications.asp

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TABLE 1—*Allele frequencies at the 13 CODIS STR loci for the North American Black population.*

Allele	D3S1358 $N^* = 7{,}602$	vWA 7,854	FGA 7,419	D8S1179 7,796	D21S11 7,652	D18S51 7,463	D5S818 7,845	D13S317 7,833	D7S820 7,612	D16S539 4,548	TH01 4,530	TPOX 4,532	CSF1PO 4,549
3	...	...	...	...	...	...	0.0001	...	...	...	...	...	...
5	...	...	...	...	...	...	...	...	...	0.0007	0.0034	...	...
6	...	...	...	...	...	...	...	...	0.0011	...	0.1283	0.0696	0.0007
7	...	...	...	...	...	...	0.0033	0.0001	0.0077	0.0002	0.4046	0.0225	0.0610
8	...	...	...	0.0024	...	0.0001	0.0555	0.0260	0.2040	0.0284	0.2214	0.3207	0.0650
9	...	...	...	0.0049	...	0.0013	0.0190	0.0218	0.1140	0.2097	0.1428	0.2308	0.0370
9.3	...	...	...	...	...	...	...	...	...	...	0.0894	...	...
10	...	...	...	0.0232	...	0.0019	0.0598	0.0273	0.3390	0.1323	0.0092	0.0966	0.2716
10.2	...	...	...	...	...	0.0016	...	...	...	...	...	...	...
11	0.0003	0.0074	...	0.0445	...	0.0050	0.2370	0.2940	0.2040	0.2902	0.0002	0.2351	0.2337
12	0.0045	0.0012	...	0.1190	...	0.0625	0.3530	0.4290	0.1080	0.1918	...	0.0235	0.2743
13	0.0077	0.0169	...	0.2030	...	0.0476	0.2510	0.1520	0.0202	0.1271	...	0.0004	0.0481
13.2	...	...	...	...	...	0.0050	...	...	...	...	...	...	...
14	0.0905	0.0632	...	0.3350	...	0.0658	0.0175	0.0486	0.0021	0.0184	...	...	0.0073
14.2	...	...	...	...	...	0.0037	...	...	...	...	...	...	...
15	0.2920	0.2130	...	0.1940	...	0.1660	0.0029	0.0010	...	0.0007	...	...	0.0007
15.2	0.0010	...	...	...	...	0.0001	...	...	...	...	...	...	...
16	0.3300	0.2600	...	0.0606	...	0.1810	0.0010	0.0002	...	...	...	...	...
17	0.2070	0.2030	0.0015	0.0119	...	0.1640	0.0002	...	...	...	...	...	...
17.2	...	...	0.0008	...	...	...	...	...	...	...	...	...	...
18	0.0630	0.1390	0.0075	0.0012	...	0.1160	...	...	...	...	...	...	...
18.2	...	...	0.0124	...	...	...	...	...	...	...	...	...	...
19	0.0048	0.0697	0.0665	0.0001	...	0.0913	...	...	...	...	...	...	...
19.2	...	...	0.0028	...	...	0.0004	...	...	...	...	...	...	...
20	...	0.0223	0.0620	...	...	0.0537	...	...	...	...	...	...	...
20.2	...	...	0.0014	...	...	...	...	...	...	...	...	...	...
21	...	0.0043	0.1120	...	...	0.0222	...	...	...	...	...	...	...
21.2	...	...	0.0015	...	...	...	...	...	...	...	...	...	...
22	...	0.0007	0.1800	...	...	0.0086	...	...	...	...	...	...	...
22.2	...	...	0.0020	...	...	...	...	...	...	...	...	...	...
23	...	...	0.1780	...	...	0.0021	...	...	...	...	...	...	...
23.2	...	...	0.0010	...	...	...	...	...	...	...	...	...	...
24	...	...	0.1680	...	...	0.0003	...	...	...	...	...	...	...
24.2	...	...	0.0001	...	...	...	...	...	...	...	...	...	...
25	...	...	0.0989	...	0.0003	0.0002	...	...	...	...	...	...	...
25.2	...	...	0.0002	...	0.0001	...	...	...	...	...	...	...	...
26	...	...	0.0506	...	0.0012	...	...	...	...	...	...	...	...
26.2	...	...	0.0001	...	...	...	...	...	...	...	...	...	...
27	...	...	0.0319	...	0.0560	...	...	...	...	...	...	...	...
28	...	...	0.0125	...	0.2430	...	...	...	...	...	...	...	...
28.2	...	...	...	...	0.0001	...	...	...	...	...	...	...	...
29	...	...	0.0050	...	0.1890	...	...	...	...	...	...	...	...
29.2	...	...	0.0001	...	0.0006	...	...	...	...	...	...	...	...
30	...	...	0.0018	...	0.1870	...	...	...	...	...	...	...	...
30.2	...	...	0.0014	...	0.0192	...	...	...	...	...	...	...	...
31	...	...	0.0047	...	0.0809	...	...	...	...	...	...	...	...
31.2	...	...	...	...	0.0520	...	...	...	...	...	...	...	...
32	...	...	...	...	0.0172	...	...	...	...	...	...	...	...
32.2	...	...	...	...	0.0694	...	...	...	...	...	...	...	...
33	...	...	...	...	0.0046	...	...	...	...	...	...	...	...
33.2	...	...	...	...	0.0308	...	...	...	...	...	...	...	...
34	...	...	...	...	0.0059	...	...	...	...	...	...	...	...
34.2	...	...	...	...	0.0031	...	...	...	...	...	...	...	...
35	...	...	...	...	0.0314	...	...	...	...	...	...	...	...
35.2	...	...	...	...	0.0003	...	...	...	...	...	...	...	...
36	...	...	...	...	0.0061	...	...	...	...	...	...	...	...
36.2	...	...	...	...	...	...	...	...	...	...	...	...	...
37	...	...	...	...	0.0014	...	...	...	...	...	...	...	...
38	...	...	...	...	0.0005	...	...	...	...	...	...	...	...
$p^\dagger$	0.188	0.019	0.148	0.512	0.871	0.451	0.180	0.508	0.665	0.981	0.137	0.544	0.588
He (%)	75.1	81.7	87.3	78.8	85.0	87.7	74.9	70.2	77.7	80.0	74.2	77.3	78.5
Ho (%)	74.6	81.9	87.3	78.8	84.6	88.1	74.4	70.3	77.4	80.5	73.7	77.0	78.9
PD	0.932	0.942	0.971	0.926	0.943	0.973	0.897	0.864	0.917	0.931	0.897	0.914	0.921
PE	0.503	0.635	0.741	0.577	0.687	0.757	0.499	0.433	0.552	0.608	0.488	0.545	0.579

* Number of individuals included in the analysis that fit the qualitative specifications described in the text; † Fisher exact test based on 2,000 shufflings; He: expected heterozygosity; Ho: observed heterozygosity; PD: Power of Discrimination; PE: Probability of Exclusion; . . . allele not detected or not applicable.

Interclass correlations yielding $p < 0.05$ for pairwise comparisons: D8S1179/D13S317, vWA/TH01, D7S820/D5S818 and FGA/TH01.

TABLE 2—*Allele frequencies at the 13 CODIS STR loci for the North American Caucasian population.*

Allele	D3S1358 $N^* = 7{,}636$	vWA 7,837	FGA 7,674	D8S1179 7,731	D21S11 7,730	D18S51 7,628	D5S818 7,829	D13S317 7,814	D7S820 7,685	D16S539 3,723	TH01 3,699	TPOX 3,701	CSF1PO 3,722
3	...	...	...	...	...	...	...	...	...	...	...	...	...
5	...	...	...	...	...	...	...	...	...	...	0.0020	...	...
6	...	...	...	...	...	...	0.0001	...	0.0001	...	0.2310	0.0011	0.0003
7	...	...	...	...	...	...	0.0020	0.0003	0.0208	0.0001	0.1879	0.0012	0.0008
8	...	...	...	0.0169	...	...	0.0033	0.1200	0.1560	0.0157	0.1077	0.5376	0.0039
9	...	...	...	0.0127	...	0.0008	0.0355	0.0754	0.1650	0.1125	0.1480	0.1038	0.0218
9.3	...	...	...	...	...	...	...	...	...	...	0.3150	...	...
10	...	...	...	0.0915	...	0.0091	0.0584	0.0618	0.2680	0.0608	0.0072	0.0561	0.2636
10.2	...	...	...	...	...	...	...	...	...	...	...	...	...
11	0.0009	0.0001	...	0.0724	...	0.0112	0.3690	0.3110	0.2040	0.2988	0.0001	0.2588	0.2937
12	0.0007	0.0004	...	0.1470	...	0.1450	0.3620	0.2830	0.1450	0.3002	...	0.0403	0.3291
13	0.0031	0.0010	...	0.3260	...	0.1260	0.1590	0.1040	0.0333	0.1825	...	0.0001	0.0689
13.2	...	...	...	...	...	0.0001	...	...	...	...	...	...	...
14	0.1240	0.0965	...	0.1980	...	0.1640	0.0093	0.0443	0.0071	0.0265	...	...	0.0149
14.2	...	...	...	...	...	...	...	...	...	...	...	...	...
15	0.2690	0.0990	...	0.1060	...	0.1440	0.0010	0.0014	0.0002	0.0017	...	...	0.0020
15.2	...	...	...	...	...	...	...	...	...	...	...	...	...
16	0.2430	0.2180	...	0.0267	...	0.1280	...	...	0.0001	...	...	...	...
17	0.2000	0.2700	0.0008	0.0036	...	0.1160	...	...	...	...	...	...	...
17.2	...	...	...	...	...	...	...	...	...	...	...	...	...
18	0.1460	0.2140	0.0162	0.0002	...	0.0773	...	...	...	...	...	...	...
18.2	...	...	...	...	...	...	...	...	...	...	...	...	...
19	0.0125	0.0847	0.0636	...	...	0.0440	...	...	...	...	...	...	...
19.2	...	...	0.0001	...	...	...	...	...	...	...	...	...	...
20	0.0001	0.0155	0.1470	...	...	0.0173	...	...	...	...	...	...	...
20.2	...	...	0.0015	...	...	...	...	...	...	...	...	...	...
21	...	0.0011	0.1770	...	...	0.0098	...	...	...	...	...	...	...
21.2	...	...	0.0032	...	...	...	...	...	...	...	...	...	...
22	...	0.0001	0.1770	...	...	0.0046	...	...	...	...	...	...	...
22.2	...	...	0.0100	...	...	...	...	...	...	...	...	...	...
23	...	...	0.1410	...	...	0.0014	...	...	...	...	...	...	...
23.2	...	...	0.0044	...	...	...	...	...	...	...	...	...	...
24	...	...	0.1340	...	...	0.0005	...	...	...	...	...	...	...
24.2	...	...	0.0011	...	0.0008	...	...	...	...	...	...	...	...
25	...	...	0.0850	...	0.0001	...	...	...	...	...	...	...	...
25.2	...	...	0.0001	...	0.0008	...	...	...	...	...	...	...	...
26	...	...	0.0300	...	0.0021	0.0001	...	...	...	...	...	...	...
26.2	...	...	0.0001	...	0.0002	...	...	...	...	...	...	...	...
27	...	...	0.0059	...	0.0332	...	...	...	...	...	...	...	...
28	...	...	0.0016	...	0.1580	...	...	...	...	...	...	...	...
28.2	...	...	...	...	0.0002	...	...	...	...	...	...	...	...
29	...	...	0.0003	...	0.2120	...	...	...	...	...	...	...	...
29.2	...	...	...	...	0.0010	...	...	...	...	...	...	...	...
30	...	...	0.0001	...	0.2580	...	...	...	...	...	...	...	...
30.2	...	...	...	...	0.0353	...	...	...	...	...	...	...	...
31	...	...	0.0001	...	0.0737	...	...	...	...	...	...	...	...
31.2	...	...	...	...	0.0896	...	...	...	...	...	...	...	...
32	...	...	...	...	0.0142	...	...	...	...	...	...	...	...
32.2	...	...	...	...	0.0854	...	...	...	...	...	...	...	...
33	...	...	...	...	0.0021	...	...	...	...	...	...	...	...
33.2	...	...	...	...	0.0290	...	...	...	...	...	...	...	...
34	...	...	...	...	0.0001	...	...	...	...	...	...	...	...
34.2	...	...	...	...	0.0035	...	...	...	...	...	...	...	...
35	...	...	...	...	0.0002	...	...	...	...	...	...	...	...
35.2	...	...	...	...	0.0004	...	...	...	...	...	...	...	...
36	...	...	...	...	0.0001	...	...	...	...	...	...	...	...
36.2	...	...	...	...	0.0001	...	...	...	...	...	...	...	...
37	...	...	...	...	...	...	...	...	...	...	...	...	...
38	...	...	...	...	...	...	...	...	...	...	...	...	...
$p^\dagger$	0.282	0.384	0.310	0.097	0.425	0.193	0.988	0.523	0.738	0.647	0.702	0.042	0.641
He (%)	79.1	80.8	86.5	80.7	83.9	87.7	70.3	78.7	81.2	77.0	77.9	62.9	73.1
Ho (%)	78.1	81.2	86.4	80.3	84.0	87.2	71.1	79.5	80.5	76.7	78.5	63.1	73.0
PD	0.923	0.903	0.967	0.939	0.956	0.972	0.860	0.925	0.938	0.912	0.916	0.812	0.879
PE	0.564	0.622	0.723	0.605	0.675	0.738	0.445	0.590	0.608	0.539	0.571	0.330	0.476

* Number of genotypes included in the analysis that fit the qualitative specifications outlined in the text; † Fisher exact test based on 2,000 shufflings; He: expected heterozygosity; Ho: observed heterozygosity; PD: Power of Discrimination; PE: Probability of Exclusion; . . . allele not detected or not applicable.

Interclass correlations yielding $p < 0.05$ for pairwise comparisons: FGA/D3S1358 and D3S1358/D21S11.

TABLE 3—*Allele frequencies at the 13 CODIS STR loci for the North American Hispanic population.*

Allele	D3S1358 $N^* = 690$	vWA 703	FGA 939	D8S1179 679	D21S11 683	D18S51 647	D5S818 705	D13S317 703	D7S820 939	D16S539 993	TH01 993	TPOX 993	CSF1PO 994
3	...	...	...	...	...	...	...	...	...	...	...	...	...
5	...	...	...	...	...	...	...	...	...	...	...	...	...
6	...	...	...	...	...	...	...	...	...	...	0.3610	0.0065	...
7	...	...	...	...	...	...	0.0482	...	0.0138	...	0.3077	0.0020	0.0055
8	...	...	...	0.0066	...	...	0.0142	0.0789	0.1283	0.0136	0.0549	0.4562	0.0065
9	...	...	...	0.0096	...	...	0.0475	0.1679	0.0863	0.1123	0.0951	0.0645	0.0211
9.3	...	...	...	...	...	...	...	...	...	...	0.1752	...	...
10	...	...	...	0.0862	...	0.0077	0.0489	0.0789	0.2662	0.2205	0.0060	0.0322	0.2420
10.2	...	...	...	...	...	0.0008	...	...	...	...	...	...	...
11	0.0022	0.0021	...	0.0611	...	0.0116	0.4064	0.2198	0.2796	0.2649	...	0.2729	0.2842
12	0.0014	0.0014	...	0.1215	...	0.1221	0.3050	0.2660	0.1837	0.2638	...	0.1636	0.3597
13	0.0051	0.0050	...	0.2946	...	0.1105	0.1234	0.1273	0.0373	0.1067	...	0.0020	0.0729
13.2	...	...	...	...	...	0.0008	...	...	...	...	...	...	...
14	0.0746	0.0626	...	0.2533	...	0.1700	0.0064	0.0597	0.0048	0.0171	...	...	0.0070
14.2	...	...	...	...	...	...	...	...	...	...	...	...	...
15	0.3659	0.1024	0.0005	0.1318	...	0.1406	...	0.0014	...	0.0010	...	...	0.0010
15.2	...	...	...	...	...	...	...	...	...	...	...	...	...
16	0.2862	0.3151	...	0.0309	...	0.1182	...	...	...	...	...	...	...
17	0.1565	0.2639	0.0016	0.0037	...	0.1708	...	...	...	...	...	...	...
17.2	...	...	...	...	...	...	...	...	...	...	...	...	...
18	0.0993	0.1664	0.0096	0.0007	...	0.0665	...	...	...	...	...	...	...
18.2	...	...	...	...	...	...	...	...	...	...	...	...	...
19	0.0080	0.0669	0.0815	...	...	0.0309	...	...	...	...	...	...	...
19.2	...	...	0.0011	...	...	...	...	...	...	...	...	...	...
20	0.0007	0.0142	0.1054	...	...	0.0193	...	...	...	...	...	...	...
20.2	...	...	...	...	...	...	...	...	...	...	...	...	...
21	...	...	0.1214	...	...	0.0116	...	...	...	...	...	...	...
21.2	...	...	...	...	...	...	...	...	...	...	...	...	...
22	...	...	0.1432	...	...	0.0131	...	...	...	...	...	...	...
22.2	...	...	0.0021	...	...	...	...	...	...	...	...	...	...
23	...	...	0.1470	...	...	0.0031	...	...	...	...	...	...	...
23.2	...	...	0.0037	...	...	...	...	...	...	...	...	...	...
24	...	...	0.1539	...	...	0.0015	...	...	...	...	...	...	...
24.2	...	...	0.0005	...	...	...	...	...	...	...	...	...	...
25	...	...	0.1315	...	...	0.0008	...	...	...	...	...	...	...
25.2	...	...	...	...	...	...	...	...	...	...	...	...	...
26	...	...	0.0650	...	0.0015	...	...	...	...	...	...	...	...
26.2	...	...	...	...	...	...	...	...	...	...	...	...	...
27	...	...	0.0229	...	0.0198	...	...	...	...	...	...	...	...
28	...	...	0.0064	...	0.1069	...	...	...	...	...	...	...	...
28.2	...	...	...	...	...	...	...	...	...	...	...	...	...
29	...	...	0.0011	...	0.2138	...	...	...	...	...	...	...	...
29.2	...	...	...	...	0.0037	...	...	...	...	...	...	...	...
30	...	...	0.0011	...	0.2562	...	...	...	...	...	...	...	...
30.2	...	...	...	...	0.0300	...	...	...	...	...	...	...	...
31	...	...	0.0005	...	0.0827	...	...	...	...	...	...	...	...
31.2	...	...	...	...	0.1171	...	...	...	...	...	...	...	...
32	...	...	...	...	0.0095	...	...	...	...	...	...	...	...
32.2	...	...	...	...	0.1061	...	...	...	...	...	...	...	...
33	...	...	...	...	0.0015	...	...	...	...	...	...	...	...
33.2	...	...	...	...	0.0425	...	...	...	...	...	...	...	...
34	...	...	...	...	0.0029	...	...	...	...	...	...	...	...
34.2	...	...	...	...	0.0007	...	...	...	...	...	...	...	...
35	...	...	...	...	0.0037	...	...	...	...	...	...	...	...
35.2	...	...	...	...	...	...	...	...	...	...	...	...	...
36	...	...	...	...	0.0007	...	...	...	...	...	...	...	...
36.2	...	...	...	...	...	...	...	...	...	...	...	...	...
37	...	...	...	...	0.0007	...	...	...	...	...	...	...	...
38	...	...	...	...	...	...	...	...	...	...	...	...	...
$p^\dagger$	0.863	0.265	0.894	0.228	0.246	0.317	0.487	0.661	0.752	0.229	0.335	0.481	0.186
He (%)	74.5	78.5	88.0	80.5	84.3	87.5	72.0	82.1	79.2	78.8	73.3	68.6	72.6
Ho (%)	73.9	78.5	88.2	80.9	81.1	87.3	70.4	78.9	78.3	77.1	71.0	68.2	70.6
PD	0.894	0.923	0.973	0.936	0.957	0.971	0.879	0.944	0.926	0.922	0.884	0.852	0.876
PE	0.491	0.571	0.758	0.616	0.620	0.741	0.435	0.580	0.568	0.546	0.444	0.401	0.438

* Number of genotypes included in the analysis that fit the qualitative specifications outlined in the text; † Fisher exact test based on 2,000 shufflings; He: expected heterozygosity; Ho: observed heterozygosity; PD: Power of Discrimination; PE: Probability of Exclusion; . . . allele not detected or not applicable.

Interclass correlations yielding $p < 0.05$ for pairwise comparisons: D3S1358/D21S11, D3S1358/D13S317, vWA/D21S11, D21S11/D13S317 and D16S539/TH01.

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