

TECHNICAL NOTE

Bruce Budowle,¹ Ph.D.; Tamyra R. Moretti,¹ Ph.D.; Anne L. Baumstark,¹ B.S.;
Debra A. Defenbaugh,¹ B.S.; and Kathleen M. Keys,¹ B.S.

Population Data on the Thirteen CODIS Core Short Tandem Repeat Loci in African Americans, U.S. Caucasians, Hispanics, Bahamians, Jamaicans, and Trinidadians*

REFERENCE: Budowle B, Moretti TR, Baumstark AL, Defenbaugh DA, Keys KM. Population data on the thirteen CODIS core short tandem repeat loci in African Americans, U.S. Caucasians, Hispanics, Bahamians, Jamaicans, and Trinidadians. *J Forensic Sci* 1999;44(6):1277–1286.

ABSTRACT: Allele distributions for 13 tetrameric short tandem repeat (STR) loci, CSF1PO, FGA, TH01, TPOX, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, and D21S11, were determined in African American, United States Caucasian, Hispanic, Bahamian, Jamaican, and Trinidadian sample populations. There was little evidence for departures from Hardy-Weinberg expectations (HWE) in any of the populations. Based on the exact test, the loci that departed significantly from HWE are: D21S11 ($p = 0.010$, Bahamians); CSF1PO ($p = 0.014$, Trinidadians); TPOX ($p = 0.011$, Jamaicans and $p = 0.035$, U.S. Caucasians); and D16S539 ($p = 0.043$, Bahamians). After employing the Bonferroni correction for the number of loci analyzed (i.e., 13 loci per database), these observations are not likely to be significant. There is little evidence for association of alleles between the loci in these databases. The allelic frequency data are similar to other comparable data within the same major population group.

KEYWORDS: forensic science, Caucasian, African American, Hispanic, Bahamian, Jamaican, Trinidadian, DNA typing, population genetics, CSF1PO, FGA, TH01, TPOX, VWA, D3S1358, D5S818, D7S820, D13S317, D16S539, D18S51, D21S11, CODIS core loci

The full use of DNA typing technology in forensic science has come to fruition by the development of national DNA databanks, such as the Combined DNA Index System (CODIS) (1). The main objective for a national DNA databank is to assist investigators in the identification of perpetrators of violent crimes. For purposes of applying DNA technology to human identity testing and to make effective use of a national DNA databank, defined polymorphic genetic

markers are required, and all laboratories that contribute to the database should use the same genetic loci. Short tandem repeat (STR) loci are the most informative PCR-based genetic markers available to date for attempting to individualize biological material (2–5). The 13 STR loci CSF1PO, FGA, TH01, TPOX, vWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, and D21S11 have been selected as the core loci for use in CODIS (1).

This paper presents allele distribution data in African Americans, United States Caucasians, Hispanics, Bahamians, Jamaicans, and Trinidadians for the 13 core STR loci. The data demonstrate that these loci can be useful for providing estimates of the frequency of a DNA profile in forensic identity testing and that a multiple locus profile is extremely rare in all the population groups analyzed.

Materials and Methods

Sample Preparation—Whole blood, obtained in EDTA vacutainer tubes by venipuncture from African American, Caucasian, and southwestern Hispanic individuals, was kindly provided by Dr. A. Eisenberg, University of North Texas Health Science Center, Fort Worth, Texas. Bloodstains from Bahamians, Jamaicans, and Trinidadians were kindly provided by Dr. George Duncan, Broward County Sheriff's Department, Ft. Lauderdale, FL. The DNA was extracted by the phenol-chloroform method (6). The quantity of extracted DNA was estimated using the slot-blot procedure described by Wayne, et al. (7) and Budowle, et al. (8).

STR Amplification—The African American, Caucasian, and Hispanic samples were amplified at the loci FGA, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D18S51, and D21S11 using the AmpF/STR™ Profiler Plus kit (PE Biosystems, Foster City, CA) (i.e., Profiler Plus kit) and at the loci CSF1PO, TPOX, TH01, D3S1358, D7S820, and D16S539 using the AmpF/STR Cofiler™ kit (PE Biosystems, Foster City, CA) (i.e., Cofiler kit). The loci D3S1358 and D7S820 were typed with both kits. The Bahamian, Jamaican, and Trinidadian samples were amplified at the loci CSF1PO, TPOX, TH01, VWA, D5S818, D7S820, D13S317, and D16S539 using the GenePrint™ PowerPlex™ kit (Promega Corporation, Madison, WI) (i.e., PowerPlex

¹ Forensic Science Research and Training Center, FBI Academy, Quantico, Virginia.

* This is publication number 99-02 of the Laboratory Division of the Federal Bureau of Investigation. Names of commercial manufacturers are provided for identification only, and inclusion does not imply endorsement by the Federal Bureau of Investigation.

Received 21 Jan. 1999; and in revised form 8 March 1999; accepted 26 March 1999.

kit) and at the loci FGA, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D18S51, and D21S11 using the AmpF/STR Profiler Plus kit (PE Biosystems, Foster City, CA). In each PCR, 1–5 ng of template DNA were used. Amplification conditions were according to the manufacturer's recommendations except for slight modifications for PowerPlex reactions (9). A Perkin Elmer GeneAmp PCR System 9600 thermal cycler was used for the PCR.

STR Typing of PowerPlex-Amplified Samples—Three μL of loading dye (10 mM NaOH, 95% formamide, 0.05% bromophenol blue, and 0.05% xylene cyanol FF) were mixed with 3 μL of PCR product. The samples were denatured for 2 min in a Perkin Elmer Model 480 DNA thermal cycler and snap cooled at 0°C , and 5 μL were loaded onto the cathodal end of the gel. The continuous, denaturing polyacrylamide gels (6%T, 5%C; bisacrylamide as the cross-linker) were 31 cm long and 0.4 mm thick and contained 7.5 M urea and 1X TBE. Gels were permitted to polymerize for a minimum of 1 h at ambient temperature. Each gel was placed in a SA 32 apparatus (Life Technologies, Gaithersburg, MD), and the electrode buffer was 1X TBE. Electrophoresis was performed initially at 80 W for approximately 5 min and then continued at 40 W at ambient temperature until the xylene cyanol tracking dye migrated to the top of the lower buffer reservoir (approximately 2–2.5 h). Without separating the glass plates, the fluorescent products were detected by scanning using an FMBIO II (Hitachi Genetic Systems, South San Francisco, CA) according to the manufacturer's recommendations. To facilitate analysis of the larger loci, the glass plates, which still house the gel, were placed back intact on the electrophoresis apparatus, and electrophoresis was continued at 40 W for approximately 1 h. Then the gel was scanned again.

STR Typing of Profiler Plus and COfiler Amplified Samples—Samples were analyzed using either the ABI PrismTM 310 Genetic

Analyzer or an ABI Prism[®] 377 DNA Sequencer (PE Biosystems, Foster City, CA) according to the manufacturer's recommended protocol.

For the ABI PrismTM 310 Genetic Analyzer, the samples were analyzed according to the manufacturer's recommendations using the separation medium Performance Optimized Polymer (POP) 4TM (PE Biosystems, Foster City, CA). For the ABI Prism 377 DNA Sequencer, the samples were analyzed according to the manufacturer's recommendations using 5% Long RangerTM gels (FMC Bioproducts, Rockland, ME) with a 36-cm well-to-read.

Statistical Analysis—Allele designations were determined by comparison of the sample fragments with those of the allelic ladders. The frequency of each allele for each locus was calculated from the numbers of each genotype in the sample set (i.e., the gene count method). Unbiased estimates of expected heterozygosity were computed as described by Edwards, et al. (3). Possible divergence from Hardy-Weinberg expectations (HWE) was tested by calculating the unbiased estimate of the expected homozygote/heterozygote frequencies (10–13) and the exact test (14), based on 2000 shuffling experiments. An interclass correlation criterion (15) for two-locus associations was used for detecting disequilibrium between the STR loci.

A $2 \times N$ contingency table exact test was used to generate a G-statistic (2000 shuffling experiments) (16,17) to test for homogeneity between sample populations. The program was kindly provided by R. Chakraborty (University of Texas School of Biomedical Sciences, Houston, Texas).

Results and Discussion

The distributions (in percent) of observed alleles for all 13 STR loci are shown in Table 1. The observed and expected homozy-

TABLE 1—Observed allele distributions (as %) for 13 STR loci in six population groups.

	African American (N=210)	Bahamian (N=157)	Jamaican (N=194)	Trinidad (N=80)	Caucasian (N=203)	Hispanic (N=209)
D3S1358						
<12	0.476	0.000	0.000	0.000	0.000	0.000
12	0.238	0.000	0.515	0.000	0.000	0.000
13	1.190	0.000	1.546	0.000	0.246	0.239
14	12.143	7.643	6.701	5.625	14.039	7.895
15	29.048	31.847	33.763	31.250	24.631	42.584
15.2	0.000	0.318	0.258	0.000	0.000	0.000
16	30.714	33.758	30.670	31.875	23.153	26.555
17	20.000	19.745	21.134	20.000	21.182	12.679
18	5.476	6.369	4.639	11.250	16.256	8.373
19	0.476	0.318	0.773	0.000	0.493	1.435
>19	0.238	0.000	0.000	0.000	0.000	0.239
Homozygosity (Obs.)	21.4%	25.5%	27.8%	16.3%	19.2%	26.3%
Homozygosity (Exp.)	23.5%	26.2%	25.8%	25.0%	20.3%	28.0%
(p)	0.482	0.838	0.513	0.070	0.691	0.595
Exact Test	0.797	0.758	0.270	0.222	0.084	0.333
PD	0.903	0.885	0.886	0.878	0.920	0.880
PE	0.543	0.499	0.508	0.511	0.589	0.492

TABLE 1—(Continued)

VWA	African American (N=180)	Bahamian (N=162)	Jamaican (N=244)	Trinidad (N=85)	Caucasian (N=196)	Hispanic (N=203)
11	0.278	0.926	0.410	0.588	0.000	0.246
13	0.556	2.778	0.820	0.588	0.510	0.000
14	6.667	6.173	7.377	8.824	10.204	6.158
15	23.611	15.123	22.746	14.118	11.224	7.635
16	26.944	26.235	29.098	29.412	20.153	35.961
17	18.333	20.679	18.238	26.471	26.276	22.167
18	13.611	18.210	13.115	13.529	22.194	19.458
19	7.222	7.099	5.328	4.706	8.418	7.143
20	2.778	2.778	2.254	1.765	1.020	1.232
21	0.000	0.000	0.615	0.000	0.000	0.000
Homozygosity (Obs.)	11.7%	17.3%	20.9%	20.0%	22.4%	24.6%
Homozygosity (Exp.)	18.9%	17.6%	19.4%	20.0%	18.7%	22.9%
(p)	0.014	0.928	0.557	0.991	0.179	0.564
Exact Test	0.328	0.790	0.655	0.229	0.063	0.928
PD	0.926	0.942	0.933	0.917	0.932	0.914
PE	0.624	0.648	0.617	0.602	0.625	0.563
FGA	African American (N=180)	Bahamian (N=157)	Jamaican (N=194)	Trinidad (N=80)	Caucasian (N=196)	Hispanic (N=203)
<18	0.278	1.274	0.773	0.000	0.000	0.000
18	0.833	0.000	0.000	1.250	3.061	0.246
18.2	0.833	1.274	2.062	0.000	0.000	0.000
19	5.278	5.732	6.701	5.625	5.612	7.882
19.2	0.278	0.000	0.773	0.000	0.000	0.000
20	7.222	7.325	4.639	9.375	14.541	7.143
20.2	0.000	0.000	0.000	0.000	0.255	0.246
21	12.500	11.146	7.474	10.000	17.347	13.054
21.2	0.000	0.318	0.000	0.000	0.000	0.246
22	22.500	14.331	18.814	16.875	18.878	17.734
22.2	0.556	0.000	0.000	0.000	1.020	0.493
22.3	0.000	0.318	0.000	0.000	0.000	0.000
23	12.500	18.153	19.588	16.250	15.816	14.039
23.2	0.000	0.000	0.000	0.000	0.000	0.739
24	18.611	20.064	14.433	20.625	13.776	12.562
24.2	0.000	0.000	0.258	0.000	0.000	0.000
24.3	0.000	0.000	0.258	0.000	0.000	0.000
25	10.000	9.554	11.598	10.625	6.888	13.793
26	3.611	3.185	4.124	4.375	1.786	8.374
27	2.222	5.096	5.155	1.875	1.020	3.202
28	1.667	0.955	1.546	1.250	0.000	0.246
29	0.556	0.637	0.773	0.625	0.000	0.000
30	0.278	0.000	0.000	0.000	0.000	0.000
>30	0.278	0.637	1.031	1.250	0.000	0.000
Homozygosity (Obs.)	12.8%	10.2%	9.3%	17.5%	12.2%	12.3%
Homozygosity (Exp.)	13.4%	12.5%	12.3%	12.8%	13.8%	12.0%
(p)	0.794	0.378	0.196	0.207	0.527	0.900
Exact Test	0.995	0.930	0.290	0.835	0.270	0.635
PD	0.964	0.967	0.962	0.962	0.959	0.969
PE	0.728	0.744	0.750	0.733	0.717	0.752

TABLE 1—(Continued)

D8S1179	African American (N=180)	Bahamian (N=157)	Jamaican (N=194)	Trinidad (N=80)	Caucasian (N=196)	Hispanic (N=203)
<9	0.278	0.000	0.000	0.625	1.786	0.246
9	0.556	0.318	0.773	0.000	1.020	0.246
10	2.500	2.229	1.289	5.000	10.204	9.360
11	3.611	5.096	3.093	7.500	5.867	6.158
12	10.833	13.057	11.340	15.625	14.541	12.069
13	22.222	18.790	20.619	23.125	33.929	32.512
14	33.333	33.758	32.216	25.000	20.153	24.631
15	21.389	18.471	21.907	17.500	10.969	11.576
16	4.444	6.051	7.990	5.625	1.276	2.463
17	0.833	2.229	0.515	0.000	0.255	0.739
18	0.000	0.000	0.258	0.000	0.000	0.000
Homozygosity (Obs.)	23.9%	24.8%	17.0%	20.0%	16.8%	18.7%
Homozygosity (Exp.)	22.0%	20.5%	21.3%	17.7%	20.1%	20.6%
(p)	0.537	0.180	0.147	0.593	0.250	0.516
Exact Test	0.696	0.168	0.284	0.896	0.786	0.061
PD	0.920	0.927	0.915	0.940	0.931	0.913
PE	0.575	0.603	0.585	0.638	0.612	0.603
D21S11	African American (N=179)	Bahamian (N=157)	Jamaican (N=194)	Trinidad (N=80)	Caucasian (N=196)	Hispanic (N=203)
24.2	0.279	0.000	0.000	0.000	0.510	0.246
24.3	0.000	0.637	0.258	0.000	0.000	0.000
26	0.279	0.000	0.000	0.000	0.000	0.000
27	6.145	7.006	6.443	6.250	4.592	0.985
28	21.508	21.975	27.320	22.500	16.582	6.897
29	18.994	17.516	18.299	20.000	18.112	20.443
29.2	0.279	0.000	0.000	0.625	0.000	0.246
30	17.877	17.834	16.495	17.500	23.214	33.005
30.2	0.838	0.955	1.804	1.250	3.827	3.202
30.3	0.000	0.318	0.000	0.000	0.000	0.000
31	9.218	9.554	6.443	5.000	7.143	6.897
31.2	7.542	5.096	4.897	8.125	9.949	8.621
32	0.838	1.911	1.546	3.125	1.531	1.232
32.1	0.000	0.000	0.258	0.000	0.000	0.000
32.2	6.983	9.873	6.186	6.875	11.224	13.547
33	0.838	0.318	0.515	0.625	0.000	0.000
33.2	3.352	3.822	3.093	5.000	3.061	4.187
34	0.838	0.637	0.773	1.875	0.000	0.000
34.2	0.279	0.318	0.258	0.000	0.000	0.493
35	2.793	2.229	4.124	1.250	0.000	0.000
35.2	0.000	0.000	0.000	0.000	0.255	0.000
36	0.559	0.000	1.031	0.000	0.000	0.000
>36	0.559	0.000	0.258	0.000	0.000	0.000
Homozygosity (Obs.)	10.6%	18.5%	14.4%	13.8%	13.3%	19.7%
Homozygosity (Exp.)	13.7%	13.7%	15.1%	13.8%	14.4%	18.7%
(p)	0.230	0.082	0.792	0.993	0.640	0.716

TABLE 1—(Continued)

Exact Test	0.495	0.010	0.917	0.912	0.415	0.655
PD	0.958	0.957	0.960	0.958	0.959	0.942
PE	0.723	0.722	0.703	0.717	0.708	0.641
D18S51	African American (N=180)	Bahamian (N=157)	Jamaican (N=194)	Trinidad (N=78)	Caucasian (N=196)	Hispanic (N=203)
<11	0.556	0.955	0.258	0.000	1.276	0.493
11	0.556	0.955	0.515	2.564	1.276	1.232
12	5.833	4.777	4.381	8.333	12.755	10.591
13	5.556	5.096	2.577	10.256	12.245	16.995
13.2	0.556	0.318	0.515	0.641	0.000	0.000
14	6.389	4.459	4.124	10.897	17.347	16.995
14.2	0.000	0.000	0.258	0.000	0.000	0.000
15	16.667	15.605	14.433	15.385	12.755	13.793
15.2	0.000	0.318	0.000	0.000	0.000	0.000
16	18.889	16.879	18.814	21.154	10.714	11.576
17	16.389	18.471	18.814	5.128	15.561	13.793
18	13.056	12.739	11.598	5.769	9.184	5.172
19	7.778	9.554	9.536	8.974	3.571	3.695
20	5.556	4.777	7.990	7.051	2.551	1.724
21	1.111	2.229	3.608	3.846	0.510	1.970
21.2	0.000	0.318	0.258	0.000	0.000	0.000
22	0.556	2.548	1.546	0.000	0.255	0.739
>22	0.556	0.000	0.773	0.000	0.000	1.232
Homozygosity (Obs.)	13.3%	11.5%	10.3%	14.1%	11.2%	11.8%
Homozygosity (Exp.)	12.5%	12.0%	12.4%	11.3%	12.2%	12.3%
(p)	0.732	0.836	0.372	0.439	0.683	0.826
Exact Test	0.920	0.263	0.573	0.523	0.622	0.512
PD	0.968	0.967	0.966	0.967	0.968	0.966
PE	0.744	0.754	0.747	0.762	0.748	0.747
D5S818	African American (N=180)	Bahamian (N=162)	Jamaican (N=244)	Trinidad (N=85)	Caucasian (N=195)	Hispanic (N=203)
7	0.278	0.000	0.000	1.176	0.000	6.158
8	5.000	7.099	5.328	2.353	0.000	0.246
9	1.389	0.926	1.025	2.941	3.077	5.419
10	6.389	5.864	5.533	15.294	4.872	6.650
11	26.111	24.383	20.492	29.412	41.026	42.118
12	35.556	37.037	39.959	32.353	35.385	29.064
13	24.444	22.531	25.615	13.529	14.615	9.606
14	0.556	1.543	1.434	2.353	0.769	0.493
15	0.000	0.617	0.615	0.000	0.256	0.246
>15	0.278	0.000	0.000	0.588	0.000	0.000
Homozygosity (Obs.)	22.2%	23.5%	29.5%	24.7%	30.3%	28.6%
Homozygosity (Exp.)	25.9%	25.4%	27.2%	23.0%	31.7%	28.1%
(p)	0.259	0.571	0.418	0.717	0.675	0.869
Exact Test	0.468	0.271	0.770	0.977	0.570	0.522
PD	0.879	0.882	0.884	0.911	0.834	0.880
PE	0.505	0.518	0.495	0.552	0.426	0.494

TABLE 1—(Continued)

D13S317	African American (N=179)	Bahamian (N=162)	Jamaican (N=244)	Trinidad (N=84)	Caucasian (N=196)	Hispanic (N=203)
7	0.000	0.309	0.205	0.000	0.000	0.000
8	3.631	2.778	2.049	5.357	9.949	6.650
9	2.793	3.086	2.459	4.762	7.653	21.921
10	5.028	2.469	2.459	5.357	5.102	10.099
11	23.743	30.556	27.664	27.976	31.888	20.197
12	48.324	39.506	45.492	32.143	30.867	21.675
13	12.570	16.049	14.344	16.071	10.969	13.793
14	3.631	5.247	5.328	8.333	3.571	5.665
15	0.279	0.000	0.000	0.000	0.000	0.000
Homozygosity (Obs.)	31.3%	34.6%	31.6%	28.6%	27.0%	17.7%
Homozygosity (Exp.)	31.0%	27.8%	30.7%	21.8%	22.7%	17.1%
(p)	0.928	0.055	0.774	0.131	0.143	0.800
Exact Test	0.298	0.173	0.051	0.068	0.395	0.993
PD	0.861	0.880	0.854	0.913	0.919	0.946
PE	0.466	0.487	0.455	0.576	0.568	0.654
D7S820	African American (N=210)	Bahamian (N=162)	Jamaican (N=244)	Trinidad (N=84)	Caucasian (N=203)	Hispanic (N=209)
6	0.000	0.000	0.410	0.000	0.246	0.239
7	0.714	1.235	0.615	0.595	1.724	2.153
8	17.381	15.123	19.877	20.833	16.256	9.809
9	15.714	12.963	13.934	11.310	14.778	4.785
10	32.381	33.642	34.426	33.333	29.064	30.622
10.1	0.000	0.000	0.205	0.000	0.000	0.000
11	22.381	21.914	18.443	22.024	20.197	28.947
11.3	0.000	0.309	0.000	0.000	0.000	0.000
12	9.048	12.346	10.246	10.119	14.039	19.139
13	1.905	2.469	1.230	1.786	2.956	3.828
14	0.476	0.000	0.615	0.000	0.739	0.478
Homozygosity (Obs.)	19.5%	23.5%	24.2%	22.6%	22.7%	23.9%
Homozygosity (Exp.)	21.7%	21.4%	22.1%	22.2%	19.2%	22.6%
(p)	0.452	0.533	0.425	0.922	0.218	0.653
Exact Test	0.490	0.159	0.257	0.364	0.321	0.422
PD	0.913	0.917	0.915	0.901	0.933	0.910
PE	0.576	0.582	0.573	0.563	0.616	0.562
CSF1PO	African American (N=210)	Bahamian (N=158)	Jamaican (N=208)	Trinidad (N=82)	Caucasian (N=203)	Hispanic (N=209)
6	0.000	0.316	0.000	0.000	0.000	0.000
7	4.286	6.329	4.808	6.707	0.246	0.239
8	8.571	5.696	6.250	5.488	0.493	0.000
9	3.333	5.063	3.125	2.439	1.970	0.718
10	27.143	23.418	27.163	27.439	25.369	25.359
10.3	0.000	0.000	0.000	0.000	0.246	0.000
11	20.476	22.152	23.317	21.341	30.049	26.555
12	30.000	28.797	29.327	27.439	32.512	39.234
13	5.476	6.962	5.288	7.927	7.143	6.459
14	0.714	0.949	0.721	1.220	1.478	0.957
15	0.000	0.316	0.000	0.000	0.493	0.478
Homozygosity (Obs.)	20.0%	16.5%	21.2%	29.3%	23.6%	30.6%
Homozygosity (Exp.)	21.7%	19.9%	22.2%	20.6%	26.4%	29.1%
(p)	0.548	0.277	0.708	0.052	0.368	0.637

TABLE 1—(Continued)

Exact Test	0.983	0.439	0.354	0.014	0.620	0.205
PD	0.921	0.925	0.909	0.917	0.872	0.861
PE	0.578	0.608	0.568	0.591	0.492	0.452
	African American (N=209)	Bahamian (N=158)	Jamaican (N=208)	Trinidad (N=82)	Caucasian (N=203)	Hispanic (N=209)
TPOX						
6	8.612	6.646	6.731	9.756	0.000	0.478
7	2.153	2.532	3.125	1.220	0.246	0.478
8	36.842	32.278	38.221	32.317	54.433	55.502
9	18.182	21.835	26.442	16.463	12.315	3.349
10	9.330	8.861	7.452	6.707	3.695	3.349
11	22.488	24.367	15.385	28.659	25.369	27.273
12	2.392	3.481	2.644	4.878	3.941	9.330
13	0.000	0.000	0.000	0.000	0.000	0.239
Homozygosity (Obs.)	21.5%	22.8%	23.6%	17.1%	33.0%	42.1%
Homozygosity (Exp.)	23.5%	22.3%	25.0%	22.5%	37.7%	39.2%
(p)	0.509	0.881	0.640	0.235	0.166	0.389
Exact Test	0.413	0.617	0.011	0.233	0.035	0.415
PD	0.907	0.914	0.892	0.895	0.793	0.791
PE	0.554	0.566	0.534	0.560	0.377	0.361
	African American (N=210)	Bahamian (N=158)	Jamaican (N=208)	Trinidad (N=82)	Caucasian (N=203)	Hispanic (N=209)
TH01						
5	0.000	0.316	0.240	0.610	0.000	0.239
6	10.952	15.190	13.942	18.293	22.660	23.206
7	44.048	37.975	35.577	31.098	17.241	33.732
8	18.571	22.785	25.481	20.732	12.562	8.134
8.3	0.000	0.000	0.000	0.000	0.246	0.000
9	14.524	12.658	15.865	20.732	16.502	10.287
9.3	10.476	9.494	8.413	7.317	30.542	24.163
10	1.429	1.582	0.481	1.220	0.246	0.239
Homozygosity (Obs.)	29.5%	24.1%	17.8%	25.6%	27.1%	22.5%
Homozygosity (Exp.)	27.1%	24.2%	24.1%	21.7%	21.5%	24.1%
(p)	0.430	0.963	0.032	0.389	0.054	0.576
Exact Test	0.137	0.911	0.116	0.392	0.423	0.910
PD	0.886	0.904	0.893	0.910	0.920	0.899
PE	0.511	0.541	0.536	0.566	0.572	0.532

TABLE 1—(Continued)

	African American (N=209)	Bahamian (N=158)	Jamaican (N=206)	Trinidad (N=82)	Caucasian (N=202)	Hispanic (N=208)
D16S539						
8	3.589	3.797	3.398	6.098	1.980	1.683
9	19.856	21.835	20.874	16.463	10.396	7.933
10	11.005	9.810	10.922	12.805	6.683	17.308
11	29.426	30.063	31.311	28.659	27.228	31.490
12	18.660	17.405	18.689	17.683	33.911	28.606
13	16.507	14.241	13.835	14.634	16.337	10.337
14	0.957	2.532	0.971	3.659	3.218	2.404
15	0.000	0.316	0.000	0.000	0.248	0.240
Homozygosity (Obs.)	18.7%	22.8%	20.9%	18.3%	20.3%	22.6%
Homozygosity (Exp.)	20.0%	19.8%	20.7%	17.8%	23.1%	22.7%
(p)	0.637	0.343	0.949	0.914	0.351	0.973
Exact Test	0.110	0.043	0.524	0.859	0.704	0.506
PD	0.923	0.930	0.923	0.938	0.909	0.908
PE	0.603	0.608	0.592	0.638	0.558	0.561

gosities, exact test for departures from HWE, discrimination probability (PD), and probability of exclusion (PE) are also provided. All loci are highly polymorphic in all sample populations with the loci TPOX in Hispanics (57.9%) and US Caucasians (67.0%) and D13S317 in Bahamians (65.4%) having the lowest observed heterozygosities, and the loci FGA in Bahamians (89.8%) and Jamaicans (90.7%), D21S11 in African Americans (89.4%) and D18S51 in Jamaicans (89.7%) displaying the highest heterozygosities. Across all sample populations, the most discriminating loci were FGA, D21S11, and D18S51. Among Caucasians and Hispanics, the TPOX locus was the least discriminating locus, while the D13S317 locus tended to be a less discriminating locus among the African-based populations. There was little evidence for departures from Hardy-Weinberg expectations (HWE) in any of the populations. Based on the exact test, the loci that departed significantly from HWE are: D21S11 ($p = 0.010$, Bahamians); CSF1PO ($p = 0.014$, Trinidadians); TPOX ($p = 0.011$, Jamaicans and $p = 0.035$, U.S. Caucasians); and D16S539 ($p = 0.043$, Bahamians). After employing the Bonferroni correction (18) for the number of loci analyzed (i.e., 13 loci per database), these observations are not likely to be significant.

An inter-class correlation test analysis was performed to detect any correlations between alleles at any of the pair-wise comparisons of the 13 loci (Table 2). For each database, there was a total of 78 pair-wise comparisons performed. The number of significant departures is at or below expected levels (5%, or 4 observations) in all groups, except Caucasians. In Caucasians, nine significant departures were observed (11.5% of the pair-wise tests). However, none of the empirical levels of significance for these nine observations were below the adjusted Bonferroni level. Furthermore, three of these correlations for the Caucasian sample are negative; a positive value might be attributed to the effects of substructure. Based on these observations, the data do not support any significant departure from independence between pairs of loci in any sample population. With little evidence of association between loci, the assumption of independence is valid, and a multiple-locus profile fre-

TABLE 2—Inter-class correlation test analyses that yielded significant departures from expectation at any of the pair-wise comparisons of the 13 loci.

Database – Locus Pair	p Value
African American – D3S1358/D13S317	0.046
Bahamian – D16S539/D5S818	0.033
Bahamian – D16S539/TPOX	0.006
Bahamian – CSF1PO/D8S1179	0.026
Bahamian – VWA/D8S1179	0.008
Jamaican – TPOX/FGA	0.018
Jamaican – VWA/D8S1179	0.034
Trinidadian – D16S539/D21S11	0.038
Trinidadian – D7S820/TH01	0.023
Trinidadian – D5S818/D18S51	0.001
Caucasian – D3S1358/FGA	0.014
Caucasian – D3S1358/D8S1179	0.019
Caucasian – VWA/D8S1179	0.037
Caucasian – D8S1179/CSF1PO	0.035
Caucasian – D8S1179/D16S539	0.031
Caucasian – D18S51/D16S539	0.048
Caucasian – D5S818/D7S820	0.032
Caucasian – D7S820/D16S539	0.024
Caucasian – TPOX/D16S539	0.034
Hispanic – D18S51/TPOX	0.048
Hispanic – D5S818/D7S820	0.015

quency can be estimated using the product rule. The most common profile frequency derived from the 13 core STR loci is less than 1 in 10 billion in all populations, and usually the estimates are substantially more rare.

Significant differences (by G-statistic) in allele frequencies between the major population databases in this study were observed at some of the loci (data not shown) and might be expected. However, the allele frequencies for the African-based populations (19) tended to be more similar to each other than to Caucasians or Hispanics. Also, Caucasian allele distributions were more similar to

those in other Caucasian databases, including other US Caucasians, Swiss, Spanish, and Italians (19–23) than to African Americans (data not shown). Consistent with these observations, the average G_{st} for the 13 STR loci in African Americans and US Caucasians is an order of magnitude less than the conservative 0.01 level recommended by the National Research Council (24). Because of these similarities in allele frequencies between the compared populations, there would be no anticipated substantial differences in DNA profile frequency estimates if another subgroup sample population were used as a reference database for the 13 STR loci. A study is underway with a larger number of data sets to further define population parameters.

Because two kits from different manufacturers were used for typing our Bahamian, Jamaican, and Trinidadian samples, an opportunity arose to compare typing results generated using different primers at the loci D5S818, D7S820, and D13S317; the primers for these loci are included in the PowerPlex and Profiler Plus kits. There were no typing discrepancies for any samples at the D5S818 and D7S820 loci. However, at the D13S317 locus, nine samples were typed as heterozygotes using the Profiler Plus kit which appeared as homozygotes using the PowerPlex kit. The Promega Corporation was contacted about the null alleles at the D13S317 locus, and the primers have been modified so that amplification of the variant alleles can be accomplished. Thus, the null allele frequency at the D13S317 locus using either kit now is extremely rare. In addition, the majority of the African American, Caucasian, and Hispanic samples were typed previously for the loci CSF1PO, TPOX, and TH01 using different primers than those in the COfiler kit (25). All typing results were concordant, except for one sample in Caucasians at the CSF1PO locus. The original typing was an 11,15, but with the COfiler primers, the type appeared as a 15,15. The TPOX and TH01 types were the same and were 8,8 and 6,9.3, respectively. The presence of the CSF1PO null allele was confirmed by comparison of the peak height and peak area of the CSF1PO 15 peak height and peak area with those of the TPOX and TH01 alleles generated using the COfiler kit. Heterozygous allele peaks at a locus generally exhibit similar peak heights and peak areas to heterozygous allele peaks at other loci, labeled with the same fluorophore, in a profile. Homozygous peak heights and peak areas generally are approximately twice those of heterozygous allele peaks. The CSF1PO 15 peak height (211 rfu) and peak area (1884 rfu) were similar to the TH01 6 peak height (250 rfu) and peak area (1576 rfu), and TH01 9.3 peak height (258 rfu) and peak area (1694 rfu), but half that of the TPOX 8 peak height (505 rfu) and peak area (3662 rfu), respectively. Such concordance studies can be used to determine whether or not a null allele occurs at any substantial frequency. These observations support the conclusion that the occurrence of null alleles is rare for the loci CSF1PO, TPOX, TH01, D5S818, D7S820, and D13S317 when using either manufacturer's kits.

Kline, et al. (26) previously typed 600 samples using both the PowerPlex kit and AmpF1STR Blue™ kit (Perkin-Elmer, Foster City, CA). Both kits contain primers to amplify the vWA locus. Only one sample was typed differently when using these kits. The vWA type generated with the PowerPlex kit was 16,19 and a 16,16 was generated with the AmpF1STR Blue kit. Based on analysis of vWA types in 1483 individuals generated using both kits, Walsh (27) estimated the vWA null allele frequency to be 6.7×10^{-4} . Thus, the presence of null alleles at the vWA locus is a rare occurrence and should not present a problem when comparing vWA profiles generated using different primer sets.

In conclusion, African American, Caucasian, Hispanic, Bahamian, Jamaican, and Trinidadian databases have been established for the loci CSF1PO, FGA, TH01, TPOX, vWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, and D21S11. All loci are highly polymorphic. The application of the product rule is valid for estimating the rarity of a multiple loci profile for these 13 loci.

References

- Budowle B, Moretti TR, Niezgoda SJ, Brown BL. CODIS and PCR-based short tandem repeat loci: Law enforcement tools. In: Second European Symposium on Human Identification 1998, Promega Corporation, Madison, Wisconsin 73–88.
- Edwards A, Civitello A, Hammond HA, Caskey CT. DNA typing and genetic mapping with trimeric and tetrameric tandem repeats. *Amer J Hum Genet* 1991;49:746–56.
- Edwards A, Hammond HA, Jin L, Caskey CT, Chakraborty R. Genetic variation at five trimeric and tetrameric repeat loci in four human population groups. *Genomics* 1992;12:241–53.
- Polymeropoulos MH, Xiao H, Rath DS, Merrill CR. Tetranucleotide repeat polymorphism at the human coagulation factor XIII A subunit gene (F13A1). *Nuc Acids Res* 1991;19:4036.
- Sullivan KM, Pope S, Gill P, Robertson JM. Automated DNA profiling by fluorescent labeling of PCR products. *PCR Meth Appl* 1992;2:34–40.
- Comey CT, Koons BW, Presley KW, Smerick JB, Sobierski CA, Stanley DM, et al. DNA extraction strategies for amplified fragment length polymorphism analysis. *J Forensic Sci* 1994;39:1254–69.
- Waye JS, Presley L, Budowle B, Shuttler GG, Fourney RM. A simple method for quantifying human genomic DNA in forensic specimen extracts. *BioTechniques* 1989;7:852–5.
- Budowle B, Baechtel FS, Comey CT, Giusti AM, Klevan L. Simple protocols for typing forensic biological evidence: chemiluminescent detection for human DNA quantitation and RFLP analyses and manual typing of PCR amplified polymorphisms. *Electrophoresis* 1995;16:1559–67.
- Moretti TR, Koons BW, Budowle B. Enhancement of PCR amplification yield and specificity using AmpliTaq Gold™ DNA polymerase. *BioTechniques* 1998;25:716–22.
- Chakraborty R, Smouse PE, Neel JV. Population amalgamation and genetic variation: observations on artificially agglomerated tribal populations of Central and South America. *Amer J Hum Genet* 1988;43:709–25.
- Chakraborty R, Fornage M, Guegue R, Boerwinkle E. Population genetics of hypervariable loci: analysis of PCR based VNTR polymorphism within a population. In: Burke T, Dolf G, Jeffreys AJ, Wolff R, editors. *DNA fingerprinting: approaches and applications*. Birkhauser Verlag, Berlin. 1991;127–43.
- Nei M, Roychoudhury AK. Sampling variances of heterozygosity and genetic distance. *Genetics* 1974;76:379–90.
- Nei M. Estimation of average heterozygosity and genetic distance from a small number of individuals. *Genetics* 1978;89:583–90.
- Guo SW, Thompson EA. Performing the exact test of Hardy-Weinberg proportion for multiple alleles. *Biometrics* 1992;48:361–72.
- Karlin S, Cameron EC, Williams PT. Sibling and parent-offspring correlation estimation with variable family size. *Proc Natl Acad Sci USA* 1981;78:2664–8.
- Roff DA, Bentzen P. The statistical analysis of mitochondrial DNA polymorphisms: χ^2 and the problem of small samples. *Mol Biol Evol* 1989;6:539–45.
- Lewontin RC, Felsenstein J. The robustness of homogeneity tests in 2 X N tables. *Biometrics* 1965;21:19–33.
- Weir BS. Multiple tests. In: *Genetic data analysis*. Sinauer Associates, Inc., Sunderland, MA, 1990;109–10.
- Lins AM, Micka KA, Sprecher CJ, Taylor JA, Bacher JW, Rabbach DR, et al. Development and population study of an eight-locus short tandem repeat (STR) multiplex system. *J Forensic Sci* 1998;43:1168–80.
- Kupferschmid TD, Calicchio T, Budowle B. Maine Caucasian population DNA database using twelve short tandem repeat loci. *J Forensic Sci* 1999;44:392–5.
- Gehrig C, Hochmeister M, Borer UV, Budowle B. Swiss Caucasian population DNA data for 13 STR loci using AmpF1STR Profiler Plus and COfiler PCR amplification kits. *Forensic Sci Int* (in press).

22. Entrala C, Lorente M, Lorente JA, Alvarez JC, Moretti T, Budowle B, et al. Fluorescent multiplex analysis of nine STR loci and the amelogenin locus: Spanish population data. *Forensic Sci Int* 1998;98:179–83.
23. Garofano L, Pizzamiglio M, Vecchio C, Lago G, Floris T, D'Errico G, et al. Italian population data on thirteen short tandem repeat loci: TH01, D21S11, D18S51, VWA, FGA, D8S1179, TPOX, CSF1PO, D16S539, D7S820, D13S317, D5S818, D3S1358. *Forensic Sci Int* 1998;97:53–60.
24. National Research Council II Report. The evaluation of forensic evidence. National Academy Press, Washington, DC, 1996.
25. Budowle B, Smerick JB, Keys KM, Moretti TR. United States population data on the multiplex short tandem repeat loci—HUMTH01, TPOX, and CSF1PO and the variable number tandem repeat locus D1S80. *J Forensic Sci* 1997;42:846–9.
26. Kline MC, Jenkins B, Rodgers S. Non-amplification of a vWA allele. *J Forensic Sci* 1998;43:250.
27. Walsh S. Commentary on Kline MC, Jenkins B, Rodgers S. Non-amplification of a vWA allele. *J Forensic Sci* 1998;43:1103–4.

Additional information and reprint requests:

Bruce Budowle
Senior Scientist
Forensic Science, FBI Academy
Quantico, VA 22135