

## FOR THE RECORD

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# Population Data for Four Population Groups from the United States for the Eleven Y-Chromosome STR Loci Recommended by SWGDAM

**POPULATIONS:** Caucasian ( $n = 1243$ ), African American ( $n = 1605$ ), Hispanic ( $n = 454$ ), and Native American ( $n = 104$ ).

**KEYWORDS:** forensic science, Y-chromosome, short tandem repeats, DNA typing, human identification, population genetics, multiplex, polymerase chain reaction, Y-STR, Y-PLEX, DYS392, DYS390, DYS385a/b, DYS393, DYS389I, DYS391, DYS389II, DYS19, DYS439, DYS438

The samples for database studies were obtained from unrelated anonymous males from the indicated population groups residing in the United States of America (USA). The samples were collected as buccal swabs, blood drawn in EDTA vacutainer tubes, or blood stain cards. The DNA was extracted either by Chelex<sup>®</sup> method (1) or by using the QIAamp<sup>®</sup> MiniKit (Qiagen, Valencia, CA) following the recommended procedures. The Y-PLEX<sup>™</sup>6, Y-PLEX<sup>™</sup>5, and Y-PLEX<sup>™</sup>12 systems (ReliaGene Technologies Inc., New Orleans, LA) were used for the amplification of extracted DNA. The PCRs and the analysis of amplified product were performed as recommended and described by the manufacturer (2–4).

Gene diversity at each locus, the number of haplotypes and haplotype diversity were calculated using a program developed by Chakraborty and Lee (<http://cgi.uc.edu/download/haplo>).

The Scientific Working Group on DNA Analysis Methods (SWGDAM) has identified a set of 11 short-tandem loci on the Y-chromosome (Y-STR) DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS438, and DYS439 for forensic DNA analysis in the USA (5). Of these, the Y-PLEX<sup>™</sup>6 system enables amplification of the seven loci DYS19, DYS389II, DYS390, DYS391, DYS393, and DYS385a/b and in combination with the Y-PLEX<sup>™</sup>5 system all 11 loci can be typed (2,3). The haplotypes for the 11 Y-STR loci recommended were generated either by using Y-PLEX<sup>™</sup>6 and Y-PLEX<sup>™</sup>5 systems or the Y-PLEX<sup>™</sup>12 system (a kit that enables simultaneous amplification of the 11 recommended Y-STR loci). A total of 4623 samples comprising Caucasian ( $n = 1826$ ), African American ( $n = 2239$ ), Hispanic ( $n = 454$ ), and Native American ( $n = 104$ )

TABLE 1—Allele frequencies of the 11 Y-STR loci in Caucasian, African American, Hispanic, and Native American population groups.

| Locus    | Allele | Caucasian<br>( $n = 1243$ ) |        | African<br>American<br>( $n = 1605$ ) |        | Hispanic<br>( $n = 454$ ) |        | Native<br>American<br>( $n = 104$ ) |        |
|----------|--------|-----------------------------|--------|---------------------------------------|--------|---------------------------|--------|-------------------------------------|--------|
|          |        | Count                       | %      | Count                                 | %      | Count                     | %      | Count                               | %      |
| DYS393   | 11     | 3                           | 0.241  | 3                                     | 0.187  | 4                         | 0.881  |                                     |        |
|          | 12     | 107                         | 8.608  | 65                                    | 4.050  | 66                        | 14.537 | 22                                  | 21.154 |
|          | 13     | 1001                        | 80.531 | 911                                   | 56.760 | 333                       | 73.348 | 65                                  | 62.500 |
|          | 14     | 111                         | 8.930  | 427                                   | 26.604 | 41                        | 9.031  | 15                                  | 14.423 |
|          | 15     | 20                          | 1.609  | 191                                   | 11.900 | 9                         | 1.982  | 2                                   | 1.923  |
|          | 16     | 1                           | 0.080  | 7                                     | 0.436  | 1                         | 0.220  |                                     |        |
|          | 17     |                             |        | 1                                     | 0.062  |                           |        |                                     |        |
| DYS19    | 12     |                             |        | 2                                     | 0.125  | 1                         | 0.221  |                                     |        |
|          | 13     | 61                          | 4.911  | 30                                    | 1.869  | 88                        | 19.469 | 22                                  | 21.154 |
|          | 14     | 860                         | 69.243 | 419                                   | 26.106 | 244                       | 53.982 | 58                                  | 55.769 |
|          | 15     | 225                         | 18.116 | 613                                   | 38.193 | 84                        | 18.584 | 17                                  | 16.346 |
|          | 16     | 74                          | 5.958  | 292                                   | 18.193 | 24                        | 5.310  | 6                                   | 5.769  |
|          | 17     | 22                          | 1.771  | 248                                   | 15.452 | 11                        | 2.434  | 1                                   | 0.962  |
| DYS389II | 18     |                             |        | 1                                     | 0.062  |                           |        |                                     |        |
|          | 25     | 1                           | 0.080  |                                       |        |                           |        |                                     |        |
|          | 27     | 15                          | 1.207  | 13                                    | 0.810  | 6                         | 1.322  | 1                                   | 0.962  |
|          | 28     | 202                         | 16.251 | 140                                   | 8.723  | 47                        | 10.352 | 17                                  | 16.346 |
|          | 29     | 597                         | 48.029 | 384                                   | 23.925 | 180                       | 39.648 | 44                                  | 42.308 |
|          | 30     | 300                         | 24.135 | 557                                   | 34.704 | 144                       | 31.718 | 36                                  | 34.615 |
|          | 31     | 107                         | 8.608  | 389                                   | 24.237 | 53                        | 11.674 | 6                                   | 5.769  |
| DYS390   | 32     | 18                          | 1.448  | 105                                   | 6.542  | 24                        | 5.286  |                                     |        |
|          | 33     | 3                           | 0.241  | 17                                    | 1.059  |                           |        |                                     |        |
|          | 20     |                             |        | 28                                    | 1.745  |                           |        |                                     |        |
|          | 21     | 19                          | 1.529  | 887                                   | 55.265 | 26                        | 5.727  | 3                                   | 2.885  |
|          | 22     | 146                         | 11.746 | 146                                   | 9.097  | 34                        | 7.489  | 1                                   | 0.962  |
|          | 23     | 320                         | 25.744 | 169                                   | 10.530 | 114                       | 25.110 | 48                                  | 46.154 |
|          | 24     | 570                         | 45.857 | 265                                   | 16.511 | 231                       | 50.881 | 37                                  | 35.577 |
|          | 25     | 177                         | 14.240 | 98                                    | 6.106  | 46                        | 10.132 | 15                                  | 14.423 |
|          | 26     | 9                           | 0.724  | 11                                    | 0.685  | 3                         | 0.661  |                                     |        |
| 27       | 2      | 0.161                       | 1      | 0.062                                 |        |                           |        |                                     |        |

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TABLE 1—Continued.

| Locus   | Allele    | Caucasian<br>(n = 1243) |        | African<br>American<br>(n = 1605) |        | Hispanic<br>(n = 454) |        | Native<br>American<br>(n = 104) |        |        |
|---------|-----------|-------------------------|--------|-----------------------------------|--------|-----------------------|--------|---------------------------------|--------|--------|
|         |           | Count                   | %      | Count                             | %      | Count                 | %      | Count                           | %      |        |
| DYS391  | 7         |                         |        |                                   |        | 1                     | 0.220  |                                 |        |        |
|         | 8         |                         |        | 2                                 | 0.125  |                       |        |                                 |        |        |
|         | 9         | 28                      | 2.253  | 24                                | 1.495  | 24                    | 5.286  | 16                              | 15.385 |        |
|         | 10        | 579                     | 46.581 | 1131                              | 70.467 | 242                   | 53.304 | 50                              | 48.077 |        |
|         | 11        | 616                     | 49.558 | 424                               | 26.417 | 181                   | 39.868 | 36                              | 34.615 |        |
|         | 12        | 19                      | 1.529  | 23                                | 1.433  | 6                     | 1.322  | 1                               | 0.962  |        |
|         | 13        | 1                       | 0.080  | 1                                 | 0.062  |                       |        | 1                               | 0.962  |        |
|         | DYS385a/b | 8                       |        |                                   | 1      | 0.031                 |        |                                 |        |        |
|         |           | 9                       | 2      | 0.080                             | 6      | 0.187                 |        |                                 |        |        |
|         |           | 10                      | 31     | 1.247                             | 9      | 0.280                 | 8      | 0.881                           | 1      | 0.481  |
|         |           | 10.2                    |        |                                   | 1      | 0.031                 |        |                                 |        |        |
|         |           | 11                      | 762    | 30.652                            | 355    | 11.059                | 223    | 24.559                          | 45     | 21.635 |
|         |           | 12                      | 139    | 5.591                             | 68     | 2.118                 | 38     | 4.185                           | 6      | 2.885  |
| 13      |           | 264                     | 10.619 | 109                               | 3.396  | 85                    | 9.361  | 13                              | 6.250  |        |
| 13.2    |           |                         |        | 5                                 | 0.156  |                       |        |                                 |        |        |
| 14      |           | 830                     | 33.387 | 447                               | 13.925 | 242                   | 26.652 | 48                              | 23.077 |        |
| 15      |           | 252                     | 10.137 | 409                               | 12.741 | 112                   | 12.335 | 42                              | 20.192 |        |
| 15.3    |           | 1                       | 0.040  |                                   |        |                       |        |                                 |        |        |
| 16      |           | 99                      | 3.982  | 613                               | 19.097 | 72                    | 7.930  | 18                              | 8.654  |        |
| 16.3    |           | 2                       | 0.080  |                                   |        | 4                     | 0.441  |                                 |        |        |
| 17      | 47        | 1.891                   | 588    | 18.318                            | 49     | 5.396                 | 22     | 10.577                          |        |        |
| 18      | 40        | 1.609                   | 391    | 12.181                            | 45     | 4.956                 | 6      | 2.885                           |        |        |
| 19      | 13        | 0.523                   | 172    | 5.358                             | 24     | 2.643                 | 4      | 1.923                           |        |        |
| 20      | 4         | 0.161                   | 27     | 0.841                             | 6      | 0.661                 | 3      | 1.442                           |        |        |
| 21      |           |                         | 9      | 0.280                             |        |                       |        |                                 |        |        |
| DYS389I | 11        | 3                       | 0.241  | 18                                | 1.121  | 7                     | 1.542  | 1                               | 0.962  |        |
|         | 12        | 238                     | 19.147 | 259                               | 16.137 | 57                    | 12.555 | 17                              | 16.346 |        |
|         | 13        | 806                     | 64.843 | 1062                              | 66.168 | 281                   | 61.894 | 70                              | 67.308 |        |
|         | 14        | 186                     | 14.964 | 261                               | 16.262 | 107                   | 23.568 | 16                              | 15.385 |        |
|         | 15        | 10                      | 0.805  | 5                                 | 0.312  | 2                     | 0.441  |                                 |        |        |
| DYS439  | 9         | 1                       | 0.080  |                                   |        |                       |        |                                 |        |        |
|         | 10        | 77                      | 6.195  | 36                                | 2.243  | 30                    | 6.608  | 2                               | 1.923  |        |
|         | 11        | 443                     | 35.640 | 492                               | 30.654 | 135                   | 29.736 | 41                              | 39.423 |        |
|         | 12        | 565                     | 45.455 | 794                               | 49.470 | 217                   | 47.797 | 50                              | 48.077 |        |
|         | 13        | 140                     | 11.263 | 257                               | 16.012 | 69                    | 15.198 | 11                              | 10.577 |        |
| DYS438  | 14        | 17                      | 1.368  | 26                                | 1.620  | 3                     | 0.661  |                                 |        |        |
|         | 8         | 2                       | 0.161  | 21                                | 1.308  | 1                     | 0.220  |                                 |        |        |
|         | 9         | 49                      | 3.942  | 25                                | 1.558  | 47                    | 10.352 | 4                               | 3.846  |        |
|         | 10        | 316                     | 25.422 | 181                               | 11.277 | 98                    | 21.586 | 22                              | 21.154 |        |
|         | 11        | 112                     | 9.010  | 998                               | 62.181 | 78                    | 17.181 | 33                              | 31.731 |        |
|         | 12        | 732                     | 58.890 | 371                               | 23.115 | 225                   | 49.559 | 42                              | 40.385 |        |
|         | 13        | 31                      | 2.494  | 9                                 | 0.561  | 5                     | 1.101  | 3                               | 2.885  |        |
| DYS392  | 14        | 1                       | 0.080  |                                   |        |                       |        | 1                               | 0.962  |        |
|         | 8         |                         |        | 1                                 | 0.062  |                       |        |                                 |        |        |
|         | 9         |                         |        | 2                                 | 0.125  | 1                     | 0.220  |                                 |        |        |
|         | 10        | 4                       | 0.322  | 6                                 | 0.374  | 2                     | 0.441  |                                 |        |        |
|         | 11        | 394                     | 31.698 | 1166                              | 72.648 | 143                   | 31.498 | 24                              | 23.077 |        |
|         | 12        | 51                      | 4.103  | 76                                | 4.735  | 20                    | 4.405  | 4                               | 3.846  |        |
|         | 13        | 684                     | 55.028 | 299                               | 18.629 | 231                   | 50.881 | 42                              | 40.385 |        |
|         | 14        | 101                     | 8.126  | 51                                | 3.178  | 33                    | 7.269  | 29                              | 27.885 |        |
|         | 15        | 9                       | 0.724  | 3                                 | 0.187  | 15                    | 3.304  | 3                               | 2.885  |        |
|         | 16        |                         |        | 1                                 | 0.062  | 7                     | 0.542  | 1                               | 0.962  |        |
| 17      |           |                         |        |                                   | 1      | 0.220                 |        |                                 |        |        |
| 18      |           |                         |        |                                   | 1      | 0.220                 |        |                                 |        |        |

TABLE 2—Observed haplotype diversity and haplotype match probability for the 11 Y-STR loci.

| Population                  | Haplotype Diversity (h) | Haplotype Match Probability |
|-----------------------------|-------------------------|-----------------------------|
| Caucasian (n = 1243)        | 0.996493                | 0.003507                    |
| African American (n = 1605) | 0.999291                | 0.000709                    |
| Hispanic (n = 454)          | 0.997851                | 0.002149                    |
| Native American (n = 104)   | 0.993465                | 0.006535                    |

TABLE 3—Observed most frequent haplotypes for the 11 Y-STR loci.

| Population Group                          | Haplotype (DYS393, DYS19, DYS389II, DYS390, DYS391, DYS385a/b, DYS389I, DYS439, DYS438, and DYS392) | n   |    |
|---|---|---|----|
| Caucasian (n = 1243)                      | 13, 14, 29, 24, 11, 11-14, 13, 12, 12, 13   | 42  |    |
|   | 13, 14, 29, 24, 10, 11-14, 13, 12, 12, 13   | 29  |    |
|   | 13, 14, 29, 23, 11, 11-14, 13, 12, 12, 13   | 26  |    |
|   | 13, 14, 28, 22, 10, 13-14, 12, 11, 10, 11   | 18  |    |
|   | 13, 14, 29, 24, 11, 11-14, 13, 11, 12, 13   | 16  |    |
|   | 13, 14, 29, 24, 11, 11-14, 13, 13, 12, 13   | 15  |    |
|   | 13, 14, 29, 24, 11, 11-15, 13, 12, 12, 13   | 14  |    |
|   | 13, 14, 29, 25, 11, 11-13, 13, 12, 12, 14   | 13  |    |
|   | 13, 14, 30, 24, 11, 11-14, 13, 12, 12, 13   | 12  |    |
|   | 13, 14, 30, 24, 11, 11-14, 14, 12, 12, 13   | 10  |    |
|   | 13, 14, 30, 24, 10, 11-14, 13, 12, 12, 13   | 10  |    |
|   | 13, 14, 29, 25, 11, 11-14, 13, 12, 12, 13   | 8   |    |
|   | 13, 14, 29, 24, 11, 11-14, 13, 11, 12, 13   | 8   |    |
|   | 13, 14, 29, 24, 10, 11-15, 13, 12, 12, 13   | 8   |    |
|   | 13, 14, 29, 23, 11, 11-14, 13, 13, 12, 13   | 8   |    |
|   | 13, 14, 29, 23, 10, 11-14, 13, 12, 12, 13   | 7   |    |
|   | African American (n = 1605)   | 13, 14, 29, 24, 11, 11-14, 13, 12, 12, 13 | 15 |
| 13, 15, 31, 21, 10, 16-17, 13, 12, 11, 11 |   | 11  |    |
| 13, 15, 31, 21, 11, 16-17, 13, 11, 11, 11 |   | 10  |    |
| 13, 14, 29, 23, 11, 11-14, 13, 12, 12, 13 |   | 9   |    |
| 13, 14, 29, 24, 10, 11-14, 13, 12, 12, 13 |   | 8   |    |
| 13, 15, 31, 21, 10, 16-17, 13, 11, 11, 11 |   | 8   |    |
| 13, 14, 28, 25, 11, 14-14, 12, 11, 11, 11 |   | 7   |    |
| 13, 17, 30, 21, 10, 17-18, 13, 12, 11, 11 |   | 7   |    |
| 13, 17, 30, 21, 10, 18-18, 13, 12, 11, 11 |   | 7   |    |
| 14, 15, 30, 21, 10, 15-16, 13, 12, 11, 11 |   | 7   |    |
| 14, 17, 30, 21, 10, 17-18, 13, 12, 11, 11 |   | 7   |    |
| 15, 17, 31, 21, 10, 17-19, 14, 12, 11, 11 |   | 7   |    |
| 14, 15, 30, 21, 10, 15-18, 13, 12, 11, 11 |   | 6   |    |
| 13, 14, 29, 24, 11, 11-14, 13, 11, 12, 13 |   | 6   |    |
| 13, 14, 29, 25, 11, 11-13, 13, 12, 12, 14 |   | 6   |    |
| Hispanic (n = 454)                        |   | 13, 14, 29, 24, 11, 11-14, 13, 12, 12, 13 | 12 |
|   |   | 13, 14, 29, 24, 11, 11-14, 13, 11, 12, 13 | 10 |
|   | 13, 13, 30, 24, 9, 13-14, 14, 10, 10, 11  | 7   |    |
|   | 13, 14, 30, 24, 11, 11-14, 13, 12, 12, 13   | 7   |    |
|   | 13, 14, 30, 24, 11, 11-14, 14, 12, 12, 13   | 5   |    |
|   | 13, 14, 29, 24, 11, 11-14, 13, 13, 12, 13   | 5   |    |
|   | 13, 14, 29, 24, 10, 11-14, 13, 12, 12, 13   | 4   |    |
|   | 13, 13, 32, 23, 10, 15-19, 13, 13, 10, 11   | 4   |    |
|   | 13, 14, 28, 22, 10, 13-14, 12, 11, 10, 11   | 3   |    |
|   | 12, 14, 29, 24, 11, 11-14, 13, 12, 12, 13   | 3   |    |
|   | 13, 14, 30, 24, 11, 11-14, 14, 11, 12, 13   | 3   |    |
|   | 13, 14, 29, 24, 11, 11-15, 13, 12, 12, 13   | 3   |    |
|   | 13, 14, 30, 25, 11, 11-14, 14, 12, 12, 13   | 3   |    |
|   | 13, 15, 29, 24, 11, 11-14, 13, 11, 12, 14   | 3   |    |
|   | Native American (n = 104)   | 13, 14, 29, 23, 11, 11-14, 13, 12, 12, 13 | 7  |
|   |   | 14, 14, 28, 25, 10, 16-17, 12, 12, 11, 14 | 4  |
|   |   | 14, 14, 28, 25, 10, 16-17, 12, 11, 11, 14 | 2  |
| 13, 14, 29, 24, 11, 11-14, 13, 11, 12, 13 |   | 2   |    |
| 13, 14, 30, 24, 10, 11-14, 14, 11, 13, 13 |   | 2   |    |
| 13, 14, 30, 24, 11, 11-14, 13, 12, 12, 13 |   | 2   |    |
| 12, 15, 29, 23, 9, 14-15, 13, 11, 10, 12  |   | 2   |    |
| 12, 15, 29, 23, 9, 15-15, 13, 11, 10, 11  |   | 2   |    |
| 12, 16, 29, 23, 9, 15-15, 13, 11, 10, 11  |   | 2   |    |
| 13, 13, 30, 23, 10, 17-18, 13, 12, 11, 14 |   | 2   |    |

were profiled for the seven Y-STR loci using the Y-PLEX™6 system. Of these, 3406 samples comprising Caucasian (n = 1243), African American (n = 1605), Hispanic (n = 454), and Native American (n = 104) were profiled for the 11 Y-STR loci recommended by SWGDAM. The analysis of the data for seven Y-STR loci typed using the Y-PLEX™6 is not included herein, because the conclusions are similar to that previously reported (2,6,7). Table 1 summarizes the allele frequency distribution for alleles in each locus in Caucasian, African American, Hispanic, and Native American population groups. In general, gene diversity was high-

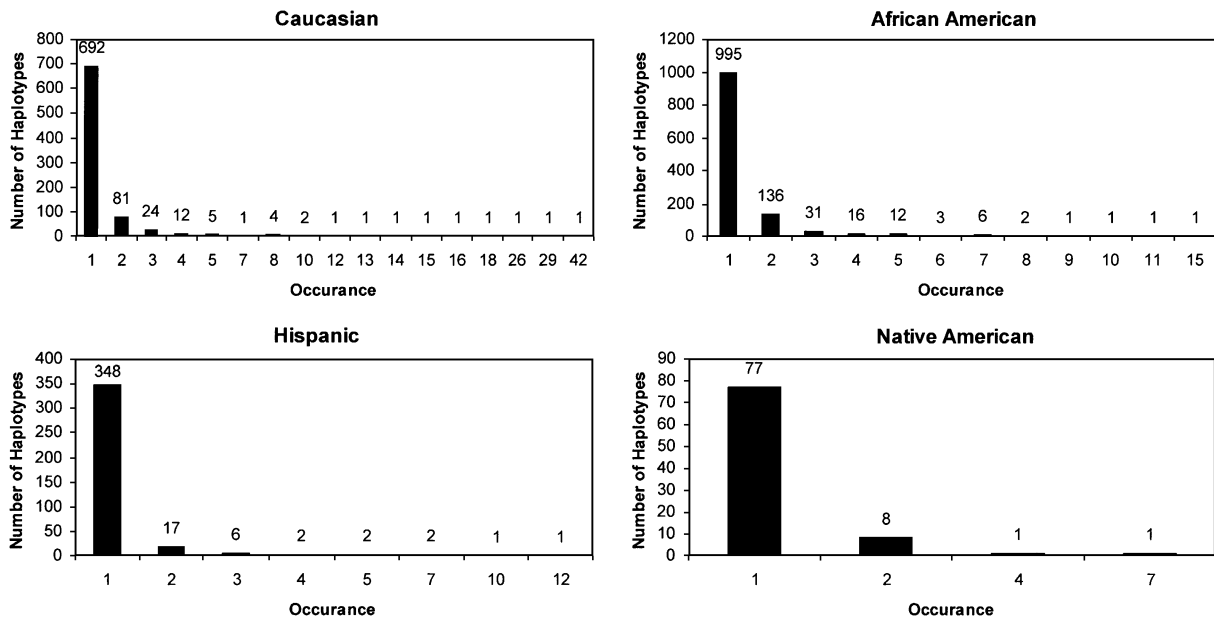


FIG. 1—Haplotype frequency distribution for 11 Y-STR loci in the Caucasian ( $n = 1243$ ), African American ( $n = 1605$ ), Hispanic ( $n = 454$ ), and Native American ( $n = 104$ ) population groups.

est for the locus DYS385a/b in all four population groups. The observed allele frequencies were similar to the database studies reported for these loci in populations of same anthropological affinity (for references see (5,6)). The haplotype genetic diversity and haplotype match probability computed for the four population groups are presented in Table 2. These values for Caucasian and African American population groups are very similar to those reported by Sinha et al. (8). The data reveal that 692 of 1243 Caucasian, 995 of 1605 African American, 348 of 454 Hispanic, and 77 of 104 Native American haplotypes were observed only once in each population group (Fig. 1). The most frequent haplotypes and their frequency distribution in Caucasian, African American, Hispanic, and Native American population groups are presented in Table 3.

Access to the data: [www.reliagene.com](http://www.reliagene.com) or via electronic mail to the corresponding author.

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