TECHNICAL NOTE

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Allele and Genotype Frequencies for D1S80 Locus in a Brazilian Population Sample*

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ABSTRACT: Gene and genotype frequencies in relation to the D1S80 locus were determined in a sample of 197 unrelated individuals (144 Caucasians and 53 Mulattoes), living in the city of São Paulo, Brazil. The Mulatto group was composed by mixed individuals who presented at least one negroid physical characteristic or declared themselves to be of mixed (Black-White) ancestry. Nineteen different alleles were detected in the Caucasian sample and 15 among Mulattoes. Alleles 18 and 24 were found to be the most common ones in the Caucasian population with frequencies of 0.173 and 0.357 respectively; the sample heterozygote frequency was estimated in 0.824. Alleles 18, 24, and 28 were found to be the most common alleles among Mulattoes with respective frequencies of 0.150, 0.349, and 0.113; the sample heterozygote frequency was 0.759. Fifty-five different genotypes were detected among Brazilian Caucasians whereas the respective figure among Mulattoes was 31. No significant deviations from Hardy-Weinberg equilibrium were found in both population samples.

KEYWORDS: forensic science, DNA typing, population genetics, D1S80, Brazil

The D1S80 locus, identified by NAKAMURA et al. (1), is very useful for forensic DNA analysis. This paper presents gene and genotype data for the D1S80 locus in a sample of 197 unrelated Brazilians individuals living in the city of São Paulo, Brazil, thus providing an useful additional database for forensic applications.

Materials and Methods

DNA was extracted from 5 mL of peripheral blood obtained from each individual of our sample by the salting-out procedure (2). PCR analysis was performed using the D1S80 Forensic DNA Amplification Reagent Kit (Perkin Elmer) under conditions recommended by the manufacturer. The amplified fragments were submitted to

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electrophoresis on a polyacrylamide gel and visualized after silver staining. Allele identification was achieved by comparison of the amplified fragments with the allelic ladder included in the reagent set. Statistical analysis: gene and genotype frequencies were estimated using standard counting procedures; for comparing gene counts between samples and for testing Hardy-Weinberg proportions within each sample, chi-squared tests were used throughout. All these procedures are described in detail by Weir (3).

Results and Discussion

Nineteen different alleles were identified in the Caucasian population and 15 were detected among Mulattoes (Table 1). Allele designation was based on the number of repeats in the core sequence.

Alleles 18 and 24 were found to be the most common alleles in the Caucasian sample, their frequencies being estimated as 0.173 and 0.357 respectively. Alleles 18, 24, and 28 were found to be the most common alleles in the Mulatto sample, with frequencies of 0.150, 0.349, and 0.113 respectively. The different genotypes detected among Brazilian Caucasians and Mulattoes from our sample are listed in Table 2.

TABLE 1—Allele frequencies at the D1S80 locus in two Brazilian ethnic

| | 0 1 | |
|--------|------------|-----------|
| Allele | Caucasians | Mulattoes |
| 17 | 0.000 | 0.019 |
| 18 | 0.173 | 0.150 |
| 20 | 0.014 | 0.019 |
| 21 | 0.031 | 0.019 |
| 22 | 0.052 | 0.057 |
| 23 | 0.014 | 0.000 |
| 24 | 0.357 | 0.349 |
| 25 | 0.028 | 0.057 |
| 26 | 0.042 | 0.019 |
| 27 | 0.014 | 0.000 |
| 28 | 0.069 | 0.113 |
| 29 | 0.062 | 0.038 |
| 30 | 0.042 | 0.019 |
| 31 | 0.056 | 0.038 |
| 32 | 0.003 | 0.000 |
| 33 | 0.003 | 0.009 |
| 34 | 0.007 | 0.009 |
| 35 | 0.007 | 0.000 |
| 36 | 0.017 | 0.009 |
| 37 | 0.007 | 0.000 |
| | | |

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TABLE 2—Distribution of genotypes at the D1S80 locus in Caucasians and Mulattoes from São Paulo, Brazil.

| | Caucasians | | Mulattoes | |
|----------------|------------|----------------|-----------|----------------|
| Genotype | No. | Frequency | No. | Frequency |
| 17,24 | _ | _ | 2 | 0.038 |
| 18,18 | 5 3 | 0.035 | _ | _ |
| 18,20 | 3 | 0.021 | _ | |
| 18,21 | 2 | 0.014 | 2 | 0.038 |
| 18,22 | 3 | 0.021 | 1 | 0.019 |
| 18,24 | 16 | 0.110 | 8 | 0.151 |
| 18,25 18,26 | 1 1 | 0.007 | 1 | 0.019 |
| 18,20 | 1 | 0.007 0.007 | _ | _ |
| 18,28 | 4 | 0.028 | 1 | 0.019 |
| 18,29 | 3 | 0.021 | _ | - |
| 18,30 | 2 | 0.014 | 1 | 0.019 |
| 18,31 | 2 | 0.014 | _ | _ |
| 18,32 | 1 | 0.007 | _ | _ |
| 18,33 | _ | _ | 1 | 0.019 |
| 18,36 | _ | | 1 | 0.019 |
| 18,37 | 1 | 0.007 | _ | _ |
| 20,24 | 1 | 0.007 | _ | |
| 20,28 | _ | _ | 1 | 0.019 |
| 20,31 | _ | _ | 1 1 | 0.019 |
| 21,21 21,22 | _ | _ | 1 | 0.019 0.019 |
| 21,24 | 3 | 0.021 | 2 | 0.019 |
| 21,24 | _ | | 1 | 0.019 |
| 21,29 | 1 | 0.007 | _ | - |
| 21,30 | _ | _ | 1 | 0.019 |
| 21,31 | _ | _ | 1 | 0.019 |
| 21,34 | 1 | 0.007 | _ | _ |
| 21,36 | 1 | 0.007 | _ | _ |
| 21,37 | 1 | 0.007 | _ | _ |
| 22,22 | 1 | 0.007 | _ | _ |
| 22,24 | 5 | 0.035 | 2 | 0.038 |
| 22,25 | 1 | 0.007 | 1 | 0.019 |
| 22,26 22,28 | 1 1 | 0.007 0.007 | 1 | 0.019 |
| 22,29 | 1 | 0.007 | | 0.019 |
| 22,30 | 1 | 0.007 | | |
| 23,24 | 4 | 0.028 | _ | _ |
| 24,24 | 15 | 0.104 | 8 | 0.151 |
| 24,25 | 1 | 0.007 | 1 | 0.019 |
| 24,26 | 6 | 0.042 | _ | _ |
| 24,27 | 2 | 0.014 | _ | _ |
| 24,28 | 8 | 0.056 | 2 | 0.038 |
| 24,29 | 10 | 0.070 | 2 | 0.038 |
| 24,30 | 5 | 0.035 | | |
| 24,31 24,34 | 8 1 | 0.056 | 1 1 | 0.019 |
| 24,34 | 1 | 0.007 0.007 | 1 | 0.019 |
| 24,36 | 2 | 0.014 | | |
| 25,25 | _ | | 1 | 0.019 |
| 25,26 | 1 | 0.007 | _ | _ |
| 25,28 | 1 | 0.007 | _ | _ |
| 25,29 | 1 | 0.007 | 1 | 0.019 |
| 25,30 | 1 | 0.007 | _ | _ |
| 25,31 | 1 | 0.007 | _ | _ |
| 26,26 | 1 | 0.007 | _ | _ |
| 26,27 | 1 | 0.007 | | |
| 26,29 | _ | _ | 1 | 0.019 |
| 26,31 28,28 | 1 | 0.007 | 1 3 | 0.019 0.057 |
| 28,30 | 1 | 0.007 | <i>3</i> | 0.037 |
| 28,31 | 1 | 0.007 | | |
| 28,33 | 1 | 0.007 | _ | _ |
| 28,35 | 1 | 0.007 | _ | _ |
| 29,29 | 1 | 0.007 | _ | _ |
| 30,30 | 1 | 0.007 | _ | _ |
| 31,31 | 1 | 0.007 | _ | _ |
| 31,36 | 2 | 0.014 | _ | |
| Total | 144 | 1.000 | 53 | 1.000 |

Fifty-five different genotypes were observed in the Caucasian population sample and 31 among Mulattoes. The most common genotypes among Brazilian Caucasians were 18/24 and 24/24, with respective frequencies of 0.111 and 0.104. Among Brazilian Mulattoes, the same genotypes were also the most frequent common, occurring in this group with equal frequencies (0.150). Most published data on D1S80 stem from studies performed with Caucasians (4-12). Our data on Brazilian Caucasians do not differ statistically from the results obtained in all these studies, in all of which 18 and 24 were found to be the most frequent alleles.

In this paper, the frequency of 11% observed for allele 28 in our Mulatto sample is straightforwardly correlated to the African background of the sample, being in agreement with Heidrich et al. (9), who described the same frequency value in Brazilian Blacks. The sample heterozygote frequency was 0.824 among Caucasians and 0.759 among Mulattoes; the corresponding figures for expected heterozigosity under panmixia were 0.822 and 0.831; no significant deviations from Hardy-Weinberg equilibrium were found in both population samples [chi-squared figures of 129.76 (171 d.f.; 0.995 > p > 0.990) and 91.86 (105 d. f.; 0.90 > p > 0.80) for Caucasians and Mulattoes respectively]. A chi-squared test for comparing gene counts between Caucasians and Mulattoes showed a non-significant value of 25.19 (19 d.f.; 0.20 > p > 0.10).

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