TECHNICAL NOTE

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Gene and Genotype Frequencies for HLA-DQA1 in Caucasians and Mulattoes in Brazil*

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ABSTRACT: Gene and genotype frequencies of the HLA-DQA1 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 consisted of mixed individuals who presented at least one negroid physical characteristic or declared themselves to be of mixed ancestry. A total of six different alleles were identified with frequencies ranging from 0.087 to 0.316 in the Caucasian population and from 0.066 to 0.330 in the Mulatto population. We observed an increased frequency of allele 1.2 among Mulattoes in relation to Caucasians. The sample heterozygote frequency was 0.722 among Caucasians and 0.736 among Mullatoes. No significant deviations from Hardy-Weinberg equilibrium were found either in the Caucasian or in the Brazilian Mullato population samples.

KEYWORDS: forensic science, DNA typing, population genetics, Brazil, HLA-DQA1, polymerase chain reaction

Gene and genotype frequencies are required for forensic DNA typing loci to evaluate probabilities of chance matches in inclusion cases. This paper presents data for the HLA-DQA1 locus in a sample of 197 unrelated Brazilian individuals living in the city of São Paulo.

Materials and Methods

DNA was extracted from 5 mL of peripheral blood obtained from each of 197 unrelated Brazilian individuals (144 Caucasians and 53 Mulattoes) by salting-out procedure (1). The amplification of the HLA-DQA1 locus and the analysis of the alleles were performed using the Amplitype HLA-DQA1 forensic DNA amplification and typing kit (Perkin Elmer) under conditions recommended by the manufacturer. 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 (2). In order to locate the categories responsible for significant values in contingency tables, the method of adjusted standardized residuals described by Haberman was applied (3,4).

Results and Discussion

Analysis of the HLA-DQA1 allele distribution in the present Brazilian population sample revealed that the most common allele was HLA-DQA1 4, present in 31% of the Caucasian population and 33% of the Mulatto population. The least frequent allele was HLA-DQA1 1.3, present in 8.7% of the Caucasian population and 6.6% of the Mulatto population. These results are similar to other population frequency studies considering Caucasian and Black populations (5–7). Different results were found in a Japanese population sample (8), in which the frequency of allele HLA-DQA1 4 was 12.3% and the frequency of allele HLA-DQA1 1.3 was 22.7%.

The other alleles HLA-DQA1 1.1; HLA-DQA1 1.2; HLA-DQA1 2 and HLA-DQA1 3 occurred at frequencies of 18.4%, 12.5%, 12.5%, and 16.3%, respectively, for the Caucasian population. For the Mulatto population their corresponding frequencies were 10.4%, 28.3%, 8.5%, and 13.2% (Table 1).

The frequency of 28.3% reported in this paper for allele HLA-DQA1 1.2 among Mulattoes certainly reflects the strong black component of this sample; studies performed in African American populations revealed similar results (9).

The most common genotypes in the Caucasian population were HLA-DQA1 3,4; HLA-DQA1 1.1,4; HLA-DQA1 2,4 and HLA-DQA1 4,4 (Table 2) and their frequencies were 12.5%, 9.0%, 9.7%, and 9.7%, respectively. The most common genotypes in the Mulatto population were HLA-DQA1 1.2,4 and HLA-DQA1 4,4. The frequencies of these genotypes were 20.7% and 9.4%, respectively. The genotypes HLA-DQA1 1.1,1.3; HLA-DQA1 1.1,2; HLA-

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TABLE 1—*Allele frequencies at the HLA–DQA1 locus in two Brazilian ethnic groups from São Paulo, Brazil.*

Alleles	Caucasians	Mulattoes
1.1	0.184	0.104
1.2	0.125	0.283
1.3	0.087	0.066
2	0.125	0.085
3	0.163	0.132
4	0.316	0.330

TABLE 2—*Distribution of genotypes at the HLA-DQA1 locus in Caucasians and Mulattoes from Säo Paulo Brazil.*

Mulattoes			Caucasians		
Genotype	No.	Frequency	Genotype	No.	Frequency
1.1, 1.1	2	0.038	1.1, 1.1	10	0.069
1.1, 1.2	2	0.038	1.1, 1.2	8	0.056
			1.1, 1.3	4	0.028
			1.1, 2	3	0.021
1.1, 3	1	0.019	1.1, 3	5	0.035
1.1, 4	4	0.075	1.1, 4	13	0.090
1.2, 1.2	4	0.075	1.2, 1.2	2	0.014
1.2, 1.3	3	0.057	1.2, 1.3	1	0.007
1.2, 2	2	0.038	1.2, 2	6	0.042
1.2, 3	4	0.075	1.2, 3	7	0.049
1.2, 4	11	0.207	1.2, 4	10	0.069
1.3, 1.3	1	0.019	1.3, 1.3	3	0.021
1.3, 2	1	0.019	1.3, 2	1	0.007
			1.3, 3	5	0.035
1.3, 4	1	0.019	1.3, 4	8	0.056
2, 2	1	0.019	2, 2	3	0.021
2, 3	1	0.019	2, 3	6	0.042
2, 4	3	0.057	2, 4	14	0.097
3, 3	1	0.019	3, 3	3	0.021
3, 4	6	0.113	3, 4	18	0.125
4, 4	5	0.094	4, 4	14	0.097
Total	53	1.000		144	1.000

DQA1 1.3,3 were not observed in the Mulatto sample. The sample heterozygote frequency was 0.722 among Caucasians and 0.736 among Mulattoes; the corresponding figures for expected heterozygosity under panmixie were 0.801 and 0.701; however, no significant deviations from Hardy-Weinberg equilibrium were found in either population sample [chi-squared values of 19.21 (15d.f.;$0.25>p>0.10$) and 12.65 (13d.f.;$0.50>p>0.25$) for Caucasians and Mulattoes respectively]. A chi-squared test for comparing gene counts between Caucasians and Mulattoes showed a significant value of 16.66 (5d.f.;$0.010>p>0.005$) that could be ascribed to an increased frequency of the 1.2 allele among Mulattoes, as shown by Haberman's test.

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References

1. Miller SA, Dykes DD, Polesky HF. A simple salting-out procedure for extracting DNA from human nucleated cells. Nucleic Acids Res 1988; 6:1215.
2. Weir BS. Genetic data analysis II. Sinauer, Sunderland, 1996.
3. Haberman SJ. The analysis of residuals in cross–classified tables. Biometrics 1973;29:205–20.
4. Everitt BS. The analysis of contingency tables. Chapman & Hall, London, 1973.
5. Sajantila A, Strom M, Budowle B, Tienari PJ, Ehnholm C, Peltonen L. The distribution of the HLA-DQα alleles and genotypes in the Finnish population as determined by the use of DNA amplification and allele specific oligonucleotides. Int J legal Med 1991;104:181–4.
6. Allen M, Saldeen T, Pettersson U, Gyllensten U. Genetic typing of HLA class II in Swedish populations: application to forensic analysis. J Forensic Sci 1993;38(3):554–70.
7. Comey CT, Budowle B. Validation studies on the analysis of the HLA-DQα locus using the polymerase chain reaction. J Forensic Sci 1991;36: 1633–48.
8. Nakajima T, Matsuki T, Ohkawara H, Nara M, Furukawa K, Kishi K. Evaluation of seven DNA markers (D1S80, HLA-DQα, LDLR, GYPA, HBGG, D7S8, and GC) in a Japanese population. Int J Legal Med 1996; 109:47–8.
9. Roche Biomedical Laboratories. Information provided in the Amplitype User guide—Perkin Elmer, 1993.

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