

FOR THE RECORD

Mercedes Fernández-Mestre,¹ Ph.D.; Miguel A. Alfonso-Sánchez,² Ph.D.; Ketevan Gendzekhadze,¹ Ph.D.; Zuly Layrisse,¹ Ph.D.; and José A. Peña,² Ph.D.

Genetic Polymorphisms at Four STR Loci from the HLA Region in a Venezuelan Population

POPULATIONS: Whole blood samples from 74 unrelated healthy individuals were collected. The donors' sample included Venezuelan mestizos from various regions of the country, but mostly from the resident population of Caracas City. A Venezuelan mestizo is the offspring of a mating between a native Venezuelan and a person born in Europe, mainly in Spain.

KEYWORDS: forensic science, DNA typing, population genetics, HLA region, TNF α , D6S265, D6S105, MICA, Venezuelan mestizo

Genomic DNA was extracted from peripheral blood according to the salting-out method (1). Ethical guidelines were adhered to as stipulated by each of the institutions involved in the study. All blood donors gave their informed consent before their inclusion in the sample. PCR amplification of *D6S105*, *D6S265*, and *TNF α* microsatellites was carried out using an end-labelled reverse oligonucleotide primer according to the methodology described in a previously published paper (2). In the case of the MICA exon 5 microsatellite, we carried out the amplification of a fragment that includes exons 2–5 by using the Expand TM Long template PCR system (Boehringer Mannheim, Indianapolis, IN). *D6S105*, *D6S265*, and *TNF α* genotyping was performed on an ABI Prism 310 Genetic Analyzer with an internal size standard (Genescan-500 TAMRA, Applied Biosystems, Foster City, CA). Allele size was determined using GENESCAN Analysis software. The sequence of MICA was determined using an ABI PRISM Dye terminator Cycle Sequencing Kit (Applied Biosystems), and the heterozygote positions were edited using the ABI sequence Navigator program.

Allelic frequencies for all the STRs analyzed in the collection of Venezuelan mestizos were estimated by the direct counting method (3). To test for Hardy–Weinberg equilibrium expectations, both the Fisher's exact *p*-method (4) and the likelihood ratio test (*G*-test) were performed. The gene diversity (GD) was calculated as described by Nei and Roychoudhury (3). The polymorphic information content (PIC) was estimated according to Botstein et al. (5), and the power of discrimination (PD) was computed as proposed by Jones (6). Genetic distances were computed with the PHYLIP program (7).

A total of 12 alleles at D6S105, 11 alleles at D6S265, nine alleles at TNF α , and five alleles at MICA exon 5 microsatellite was

observed among Venezuelan mestizos (Table 1). No significant departure from Hardy–Weinberg equilibrium expectations was detected at any of these loci. For D6S105 microsatellite, a trimodal distribution pattern seems to be clear. The most common alleles were *D6S105*6*, *D6S105*11*, and *D6S105*12*. Allele *D6S105*12* occurs at high frequencies in the African American population (8) but occurs at much lower frequencies in European populations. At the D6S265 locus, alleles 5 and 12 were found to be predominant. At the TNF α locus, a trimodal distribution pattern of alleles 2, 6, and 10 was observed. Finally, MICA showed a

TABLE 1—Allelic frequencies for STRs of the HLA region in Venezuelan mestizos.

Allele Number	D6S105* (n = 142)	D6S265† (n = 146)	TNF α ‡ (n = 148)	MICA (n = 140)
1	0.0140	0.0000	0.0337	—
2	0.0211	0.0000	0.1621	—
3	0.0211	0.0479	0.0000	—
4	0.0563	0.0068	0.0675	0.1000
5	0.1056	0.3424	0.0878	0.1500
5.1	—	—	—	0.2000
6	0.3661	0.1301	0.2702	0.2000
7	0.0915	0.2328	0.0945	—
8	0.0704	0.1164	0.0000	—
9	0.0211	0.0136	0.0000	0.3428
10	0.0070	0.0068	0.2297	—
11	0.1126	0.0479	0.0405	—
12	0.1126	0.0205	0.0000	—
13	0.0000	0.0000	0.0135	—
14	—	0.0342	0.0000	—
H _o	0.86	0.84	0.70	0.65
GD	0.81	0.79	0.82	0.77
PIC	0.79	0.77	0.80	0.73
PD	0.94	0.92	0.93	0.90

*Alleles decrease in size by 2 bp from allele 1 = 139 bp.

†Alleles increase in size by 2 bp from allele 1 = 119 bp.

‡Allele increase in size by 2 bp from allele 1 = 97 bp.

n, number of chromosomes; H_o, observed heterozygosity; GD, gene diversity; PIC, polymorphic information content; PD, power of discrimination.

¹Laboratorio de Fisiopatología, Centro de Medicina Experimental "Miguel Layrisse," Instituto Venezolano de Investigaciones Científicas, Caracas, Venezuela.

²Departamento de Genética, Antropología Física y Fisiología Animal, Universidad del País Vasco, 48080 Bilbao, Spain.

bimodal distribution pattern of alleles 6 and 9. GD values ranged from 0.77 at MICA to 0.82 at TNF α . Likewise, PIC oscillated from 0.73 at MICA to 0.80 at TNF α , whereas PD varied from 0.90 at MICA to 0.94 at D6S105.

Population data concerning the allelic frequencies of the microsatellites examined herein are yet very limited, so the information available to date mostly comes from the TNF α locus. We therefore compiled TNF α data from populations worldwide (2,9–11) in order to compute F_{ST} unbiased genetic distances (12) between all pairs of populations included (data not shown). The resultant distance matrix showed that Venezuelans displayed a relatively small genetic distance to Spaniards. Interestingly, Venezuelans appeared more closely related to Africans than to Americans, which strongly suggests the influence of the African slave trade in the genetic make-up of the contemporary Venezuelan population.

The complete dataset can be accessed via electronic mail from the communicating author: mfernand@ivic.ve

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Additional information and reprint requests:

José A. Peña, Ph.D.
Departamento de Genética
Antropología Física y Fisiología Animal
Universidad del País Vasco
Apartado 644
48080 Bilbao
Spain
E-mail: joseangel.pena@ehu.es