The Genetic Structure Of Four Argentine Ethnic Groups Reflected By The Analyses Of Ten Strs .

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Introduction:

PCR tehniques have a distinct advantage over restriction fragment length polymorphism techniques due to their ability to analyze small amounts of partially degraded samples. These features broadened the field of forensic science with special references to personal and evidentiary material identification by means of DNA typing.

Beyond its aplicability in the field of gene mapping, hereditary diseases diagnosis, forensic investigation, human remains identification in mass disasters and paternity testing, the STR systems may offer a rapid and economic approach to characterize and compare the genetic structure of populations of different ethnic origins. The availability of a high number of STR systems allows a more complete molecular insight of the groups providing results that may contribute to a more accurate anthropological interpretation that could shed light on controversial topics such as migration.

The aim of this present study is to characterize four different ethnic groups from Argentina by means ot ten STR systems, two of them located on sex chromosomes and two on autosomal chromosomes. The information obtained allowed us to compare their genetic parameters and, as a by-product, to generate a local database to be used in forensic case-work.

Materials and Methods

Populations

Blood samples were obtained from unrelated donors belonging to the followings groups: 1) Caucasoid group: Buenos Aires City Metropolitan area population (average samples size n=175); 2)Aboriginal groups: a-Mapuches (n=55) Cerro Policía, SW Province of Río Negro , b-Wichis (n=35) Santa Victoria Este, NE Province of Salta and c-Tehuelches (n=28) Chalía, SW Province of Chubut. DNA was extracted by using a CTAB-based protocol (Corach, 1992, 1994).

PCR amplification of STR loci

PCR amplifications were performed in simplex or multiplex reactions. *Loci* D6S366 (Panzer,1993), Y27H39 (Roewer,1992), HUMRENA4 (Edwards,1992), HUMCSFPO (Hammond,1994) and HUMF13A (Polymeropoulos 1991b) were amplified in simplex reactions, HUMFES/FPS (Polymeropoulos,1991a) / HUMVWA (Kimpton,1992), were amplified as duplex and HUMTHO-1 (Edwards,1992) / HUMHPRTB (Edwards,1992) / HUMFABP (Polymeropoulos,1991c) as triplex. Amplifications were performed with a Perkin Elmer Thermal Cycler. Reactions included 0.02 μCi/sample α-dATP P^{32} .

Amplicon detection

Amplification products were separated by electrophoresis through a 5% denaturing polyacrylamide sequencing gel. Electrophoresis was performed at 1500 V(constant voltage) in 1X TBE for three hours. Gels were then exposed to radiographic film.

Statistic analyses

Allele frecuency distributions and observed heterozigosity were established by counting, discriminative power and exclusion power were then calculated. In order to determine if the different groups fulfill the Hardy-Weinberg expectations, the test for H-W equilibrium was

conducted by using a chi square test (Edwards,1992; Smouse,1986). For the investigation of the X-chromosome specific STR, HUMHPRTB only female samples were considered.

Results and Discussion

Data evaluation showed differential atributtes between the metropolitan area population, with a free gene flow, and the aboriginal groups, geographically isolated and with a restricted gene flow. Hardy-Weimberg equilibrium, evaluated by comparison of the observed and expected heterozygotes, denoted deviations in the HUMFES/FPS in Metropolitans, HUMHPRTB in Mapuches and Tehuelches, and HUMF13A1 in Tehuelches. Heterozigocity defficiency was observed in HUMFES/FPS-Tehuelches, HUMFABP-Mapuches and HUMTHO-1-Wichis and can be attributed to high endogamy levels.

Both caucasoid and the three aboriginal groups displayed differences in their allele distributions as may be seen in the histograms depicted in "Fig. 1". *Loci* HUMRENA4 and HUMFABP exhibited similar allele distribution and almost identical frequency for the most common alleles in the four populations. In contrast, at other *loci* the predominant alleles differ between populations as HUMTHO-1, HUMvWA, HUMHPRTB and Y27H39. In Wichis group, only one allele appears in the Y27H39 system (variant"A"). Table I shows the most common genotypes for all nine loci combined (P) and the probability of a match (P2).

The power of exclusion (PE) and discriminative power (DP) were calculated for each *locus* in the four populations. The PE ranged from 10.6% to 64.5%, the DP from 58.8% to 94%. The STR D6S366 showed the highest PE and DP in the three groups (Metropolitan, Mapuches and Tehuelches). Instead HUMF13A1 showed the highest PE and DP in the Wichis group.

TABLE I: MATCHING PROBABILITY of STR Loci

	Met	Map	Teh	Wich
THO-1	0,17	0,28	0,25	0,5
FABP	0,31	0,48	0,50	0,35
VWA	0,14	0,29	0,18	0,28
RENA4	0,61	0,54	0,76	0,51
FES/FPS	0,21	0,27	0,5	0,25
D6S366	0,14	0,11	0,25	0,28
HPRTB	0,16	0,26	0,36	0,33
CSF1PO	0,2	0,23	0,28	0,32
F13A1	0,19	0,17	0,15	0,23
P	8,04E-07	6,35E-06	3,23E-05	4,24E-05
P2	6,47E-13	4,03E-11	1,04E-09	1,80E-09

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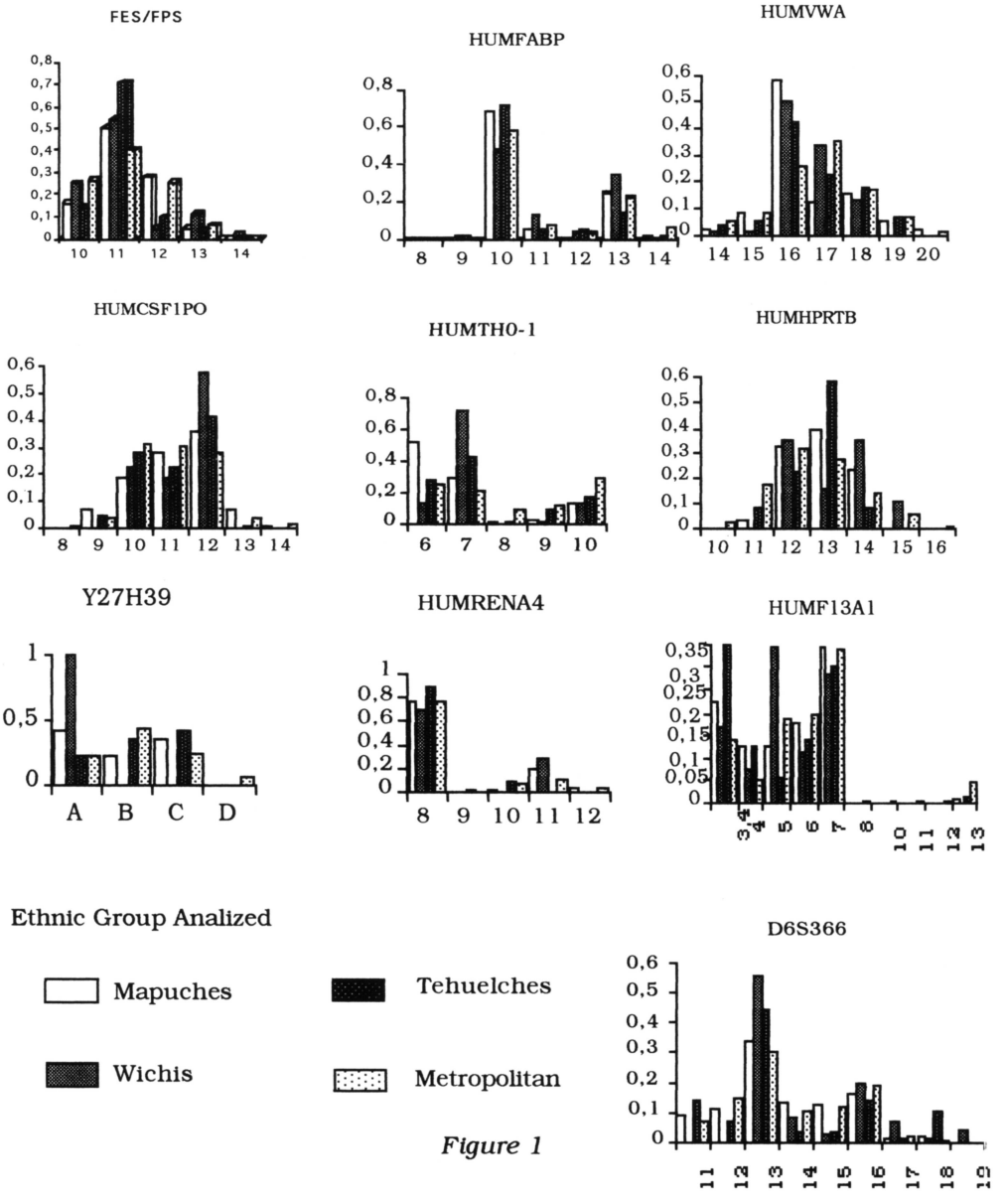

Figure 1