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Genetic variability at eleven STR loci and mtDNA in NOA populations (Puna and Calchaqui Valleys)

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Abstract. A total of 161 individuals from North Western Argentina (106 from Puna and 55 from Calchaqui Valleys) were analyzed for the mtDNA (HVRI) and for 11 autosomal STRs. The results from the STRs indicated Amerindian-European admixture in these populations, whereas the mtDNA did not show any indication of European female contributions.

Keywords: HVRI mtDNA; STRs; Puna; Calchaqui Valleys; Argentina populations

1. Introduction

Human populations from the Andean region of North Western Argentina (NOA), due to their origin and their historical-demographic peculiarities, constitute an anthropologically interesting subject for study. There is little reliable information on the structure of these populations before contact with Europeans in the late 15th century. In addition, the lack of historical data for the post-contact period means that the exact origin and/or degree of admixture of the inhabitants of this region are also unknown.

In this Andean region, two zones can be differentiated: the Puna and the Calchaqui Valleys. The Puna, in the Province of Salta, is a typical Andean plateau at high altitude. The populations in this region have extreme life conditions: low temperatures, low oxygen pressure and poor soils. In addition, they are distanced from other urban populations by poor and difficult roads. All these factors cause the isolation of these populations. The settlement model in the Puna region is dispersed with a small population density. San

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Antonio de los Cobres (altitude 3880 m) is the most populated locality (approx. 3000 inhabitants). The Calchaqui Valleys are in the East of the Puna with an altitude between 1700 and 3000 m, occupying an extension band of approximately 200 km in a north–south direction (provinces of Salta, Tucumán and Catamarca). In the pre-Hispanic epoch, these valleys were inhabited by the *diaguitas* and these pre-Hispanic societies reached the highest socioeconomic and cultural levels. The population dynamics of this zone is complex, as a consequence of the invasion of the Incas, the European colonization and, finally, the policy of estrangement of the rebels, from the XVIth until the end of the XVIIth century, which led to the disappearance of an important part of the population. The current population (25,000 inhabitants in total) has a low density and is unequally distributed, with the most populated localities being Cafayate (approx. 9000 inhabitants) and Cachi (6000). This area is a region of specific ecological and cultural characteristics, a combination of Andean and Amazonian, because it was formed with peoples from both high and low lands.

Some demographic data have been published for these populations but there is very little genetic data. The purposes of the present study were: (i) to study the STR variation in Puna and Calchaqui Valleys, and (ii) to develop a mtDNA database (HVRI) from NOA individuals.

2. Material and methods

DNA samples from 161 unrelated individuals were analyzed: 106 from Puna and 55 from Calchaqui Valleys. The STRs studied were: D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D7S820 (AmpFℓSTR Profiler Plus, Applied Biosystems), HUMF13A1 and D12S391. The HVRI mtDNA region (15997–16400) was analyzed using the QIAquick PCR purification kit (Quiagen) and the Big Dye Terminator Ready Reaction kit (Applied Biosystems). A GeneAmp PCR System 2400 and an ABI

Table 1
Statistical parameters for the STRs studied in 161 individuals from Puna (PN) and Calchaqui Valleys (CV) populations (PD: power of discrimination; PE: probability of exclusion; PE2: probability of exclusion if only one parent and child are typed)

System	Number of alleles		Heterozygosity obs./exp.		PD		PE		PE2	
	PN	CV	PN	CV	PN	CV	PN	CV	PN	CV
D3S1358	7	5	0.600/0.633	0.655/0.592	0.797	0.766	0.363	0.329	0.212	0.182
vWA	8	6	0.629/0.693	0.691/0.674	0.855	0.953	0.452	0.413	0.281	0.254
FGA	16	13	0.827/0.862	0.815/0.886	0.968	0.972	0.731	0.753	0.575	0.601
D8S1179	10	8	0.778/0.808	0.722/0.828	0.934	0.943	0.620	0.641	0.444	0.467
D21S11	13	11	0.796/0.843	0.778/0.846	0.954	0.953	0.681	0.677	0.513	0.509
D18S51	14	10	0.837/0.876	0.804/0.858	0.970	0.960	0.742	0.702	0.587	0.537
D5S818	7	7	0.613/0.723	0.611/0.708	0.886	0.867	0.503	0.469	0.321	0.293
D13S317	7	7	0.750/0.794	0.704/0.809	0.926	0.931	0.594	0.607	0.415	0.429
D7S820	8	5	0.728/0.705	0.604/0.683	0.862	0.835	0.460	0.417	0.289	0.256
F13A1	6	6	0.711/0.713	0.636/0.694	0.881	0.863	0.493	0.463	0.310	0.286
D12S391	9	9	0.738/0.737	0.691/0.718	0.883	0.875	0.497	0.487	0.323	0.314
Average	9.5	7.9	0.728/0.762	0.701/0.754	1/7.700 E+11	1/1.224 E+12	99.992 E−2	99.989 E−2	99.645 E−2	99.566 E−2

Table 2
Statistical parameters for HVRI mtDNA in Andean individuals

	Puna	Calchaqui Valleys	Total
Number of individuals	63	36	99
Number of different haplotypes	22	15	33
Number of variable sites	34	31	50
Number of unique haplotypes	10	6	13
Mean pairwise differences	4.868 ± 2.406	7.187 ± 3.449	6.355 ± 3.038
Gene diversity	0.921 ± 0.020	0.921 ± 0.026	0.952 ± 0.009
Probability of genetic identity	0.094	0.105	0.058

310 automatic sequencer (Applied Biosystems) were used. The RFLP motif −7025 *AluI* was also determined. In non-H samples, every control region sequence was assigned to a haplogroup by using the sequence motifs indicated by Richards et al. [1].

Statistical analyses were carried out with the GENEPOP [2] and ARLEQUIN packages [3]. Furthermore, the forensic parameters PD [4], PE [5] and PE2 [6] were calculated.

3. Results and discussion

The eleven STRs studied showed a combined power of discrimination (PD) of 1 in 8.424E+11 individuals and a combined probability of exclusion (PE) of 0.999925. Even when only one parent and child were typed the PE was 0.996712 (Table 1). The number of alleles observed, ranging from 7 to 17 (average 10.2), was high, taking into account that they are small and inbred populations, probably due to their Amerindian-European admixture. In mtDNA (HVRI), a total of 33 different haplotypes in 99 individuals were observed (Table 2), defined by 50 variable positions. The incidence of unique haplotypes (13.1%) was very low. In relation to shared haplotypes, 20 were shared by two or more individuals, 16 within the same population, and 4 between both populations. The gene diversity was approximately 0.92, in both populations, and the random match probability was 10%. Amazingly, the haplogroup analysis showed that all the individuals in both NOA populations had Amerindian haplogroups (A, B, C, D). Consequently, there is no indication of European female contributions for these populations. Genetic diversity of the Y-chromosome should be studied in order to estimate the proportion of Amerindian and European genes, and asymmetrical mating according to sex and ethnic group, in NOA populations.

References

[1] M. Richards, et al., Tracing European founder lineages in the near eastern mtDNA pool, Am. J. Hum. Genet. 67 (2000) 1251–1276.
[2] M. Raymond, F. Rousset, GENEPOP (version 1.2): population genetics software for exact tests and ecumenicism, J. Heredity 86 (1995) 248–249.
[3] S. Schneider, D. Roessli, L. Excoffier, ARLEQUIN ver. 2000. A Software for Population Genetics Data Analysis, Genetics and Biometry Laboratory, University of Geneva, Switzerland, 2000.
[4] R. Fisher, Standard calculations for evaluating a blood system, Heredity 5 (1951) 95–102.
[5] R. Chakraborty, L. Jin, Determination of relatedness between individuals using DNA fingerprinting, Hum. Biol. 65 (1993) 875–895.
[6] A. Chakravarti, C.C. Li, The effect of linkage on paternity calculation, in: R.H. Walker (Ed.), Inclusion Probabilities in Parentage Testing, American Association of Blood Banks, Arlington, 1983, pp. 411–422.