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Ethnic origin of y-chromosome haplotypes in subpopulations located at different altitudes in northwest Argentina

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RESUMEN

Se analizó sobre la base del polimorfismo del cromosoma Y del ADN, el origen étnico de linaje paterno en dos subpoblaciones amerindias del nordeste argentino exindidas de un mismo ancestro. Una de las subpoblaciones fue obtenida en San Salvador de Jujuy ubicado a 1.200 m sobre el nivel del mar y la segunda en los habitantes de Humahuaca con una altitud que fluctúa entre 2.500 a 3.500 metros. Encontramos un porcentaje de 40,5% de integración del cromosoma Y de origen hispano en el total de muestras de amerindios. Sin embargo esta integración fue de un 64,3% en SS de Jujuy (1.200 m aproximadamente) que es el nivel andino de menor altura y de 27.6% en Humahuaca (2.500 m o más sobre el nivel del mar) definiendo una menor integración hispánica en concordancia con la altura. ($P< 0.05$). La subpoblación que habita a 1.200 m mostró también una variación genética del cromosoma Y significativamente alta. Estos hallazgos, tienen una buena correlación con la información histórica de la conquista de América, que fue hecha por hombres que mantenían uniones poligámicas con mujeres amerindias. SS de Jujuy es la región más noreste donde se encuentra una mezcla genética de amerindios, en Argentina.

Introduction

The development of population genetics has stimulated studies on physical anthropology, giving new insights into human diversity. It has become increasingly clear that any human population, in a given territory, shares a common gene pool that may differ from those of other populations in the frequency of some of its components. The first example of a character showing variable population frequencies was that of the ABO blood groups. Nowadays, the analysis of genetic variability can be carried out at the level of the hereditary material itself (DNA), and a large number of variant markers are now available. From a biological point of view, the ethnic background of a population is usually a reflection of the frequency of certain characters genetically determined. In the case of admixed populations, the ethnic origin can be inferred by the proportion of characters contributed by the respective ancestral pools (Chakraborty et al.,1989).

Mitochondrial and Y-specific DNA do not undergo recombination and are transmitted via maternal or paternal lineages, respectively, over generations. Thus, independently from the number of generations, mutations are the only source of variation between the mtDNA and the Y-specific DNA of a given ancestor and all his/her offspring. Due to these peculiarities, mt- and Y-specific DNA polymorphic markers are extensively used to analyze the origin and evolution of human populations.

The association of two or more mitochondrial DNA (mtDNA) markers, or two or more Y-chromosome- specific DNA markers, identifies haplogroups which usually correlate with the ethnic origin of female and male ancestors (Bianchi et al., 1995, 1997; Cann et al., 1987; Pena et al., 1995; Underhill et al., 1997). Recently, Bianchi et al. (1997) used Y-chromosome-specific DNA variants to estimate the contribution of European and African Y-chromosomes to the pool of Y-chromosomes in various Amerindian populations. Moreover, the analysis of mtDNA and Y-chromosome haplotypes allowed determination of the frequency of African, Amerindian and European ancestors in male and female lineages of an African Uruguayan population as well as the asymmetric introgression of European Y-chromosomes into the ancestral population

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(Bravi et al., 1997; Sans et al., 1997). In this study, the ethnic characteristic of highland populations of the Province of Jujuy was evaluated using information from Y-specific DNA marker variation.

The province of Jujuy in the Northwest of Argentina shows an altitudinal gradient with four well defined ecological regions. Puna and Ramal correspond to the areas of highest (4000 mt.) and lowest (300 mt.) altitude respectively; Quebrada (2500 mt.) and Valle (1200 mt.) are the other two regions completing the altitudinal gradient. Human populations in these regions are of Amerindian ancestry and employ a mosaic of languages and dialects (Kunza, Quechua, Kakan, etc.) illustrating the multiethnic Amerindian origin (Atacama, Omaguaca, etc.) of the present autochtonous populations. After the Spanish conquest, the original populations received a significant Spanish input and some Negroid influence (Dipierri et al., 1997).

More recently, at the beginning of the 20th century, the admixture process of Northwest Argentina was completed by the impact of an additional immigration which shaped the present population. Recent advances in archaeological, genetics (Dipierri et al., 1998a) and ethno-historical studies, and data on settlements from national censuses seem to indicate that high altitude regions of the Province of Jujuy (Puna and "Quebrada de Humahuaca") were partially protected from the ethnic disruption which took place in Northwest Argentina during the Spanish colonial period (Dipierri et al., 1997).

Population and methods

DNA was extracted from blood collected from 42 males with surnames and phenotypes characteristic of Amerindians from the Province of Jujuy (Northwest Argentina). Fourteen samples were taken from individuals inhabiting "San Salvador de Jujuy", located at 1258 m above sea level (Valle region) and twenty-eight from donors inhabiting the "Quebrada de Humahuaca" and surrounding areas at 2500- to 3500 m aboye sea level.

Y-chromosome- specific haplotypes were identified in all males using ah (Santos et al., 1996), DYS 199 (Underhill et al., 1996), DYS19 (Santos et al., ; 995), YAP (Hammer, 1994), and pSRY (Bianchi et al., 1997) polymorphic markers.

Results and discussion

Santos et al. (1995) described a polymorphism in the alphoid subunit of the contromere of the human Y - chromosome (ah). The molecular basis of this polymorphism consists of the amplification of DNA molecules with slightly different sequences. The ah variants can be detected by PCR using a pair of primers that co-amplify two or more loci located at both edges of the Y-chromosome- specific alphoid DNA (Santos et al., 1995, 1996). Twenty-seven (h types identified as (h Ito XXVII have been detected so far (Santos et al., 1996; Bianchi and Bailliet, unpublished data, 1998). A C>T transition at bp 181 defines the two alleles of the DYS 199 locus Underhill et al., 1996). The DYS 199 allele occurs exclusively in American populations, in Siberian Eskimos, Chukchi and Even, while it was not found in individuals coming from Africa, Asia, Oceania or Europe (Underhill et al., 1996; Rodríguez-Delfín et al., 1997; Karafet et al., 1997). The presence of the T allele in Navajo and Eskimo populations suggested that the mutation probably occurred before the migration of Paleo-Indians into Central and South America (Underhill et al., 1996). DYS19 is a Y-chromosome-specific tetranucleotide microsatellite with eight different allelic forms: null, A to F., Z, and Y (Santos et al., 1995); Ciminelli et al., 1995).

The association of ahII with DYS199T and DYS19A alleles defines a Y-chromosome haplotype found in more than 80% of Amerindians (Pena et al., 1995; Bianchi et al., 1997), about 50% of Nadenes (Underhill et al., 1996; Bianchi et al., unplublished data, 1997), and likely also in Aleut-Eskimos (this ethnic group shows an association of DYS199T and DYS19A; the ah system has not been yet tested in this group). Moreover, Pena et al. (1995) analysed 46 individuals from Mongolia but they did not find any case of haplotype IIA among the 17 different Y-chromosome haplotypes detected (DYS 199T has not been yet tested in this group). On the other hand, gomolka et al. (1994) found only one case of an A allele in the DYS19 locus among 215 Asians from eight populations. So far, no other geographic population has shown the combinations of markers detailed (Pena et al., 1995; Bianchi et al., 1997; Bravi et al., 1997). As shown in Table 1, 3 out of 14 (21.4%) samples from «San Salvador de Jujuy» and 16 out of 28 (57.1%) samples from «Quebrada de Humahuaca» had this haplotype.

We now have evidence (Catanesi et al., personal communication, 1997) that haplotypes ahIIDYS199/DYS 19B, ahll/DYS 199T/DYS 19Z, and ahI/DYS199T/DYS19A (Table 1) are derived from the native American-specific haplotype (Santos et

al.,1996). Accordingly, 64.3% of samples from "Quebrada de Humahuaca" and 28.5% of samples from "San Salvador de Jujuy" had native American haplotypes (Table 1), this difference proving to be significant ($p<0.0S$, Fisher's test).

A C>T transition at the 373 bp position of the pSRY gene defines two alleles. The pSRYT variant in association with ah II and DYS19B is characteristic of European ancestry (Bianchi et al., 1997). One of our cases (Table 1) showed this haplotype. Moreover, the pSRYC/ahIUDYS 199C/ DYS19B or DYS19E haplotypes are almost exclusively detected in European Y-chromosomes. In addition to Europeans, this haplotype has also been detected at low frequency (3%) in native Mongolians (Pena et al., 1995; Santos et al., 1996; Bianchi et al., 1997; Bravi et al., 1997). From this analysis we can estimate that 57.1% of Y-chromosomes from "San Salvador de Jujuy" and 32.1% from "Quebrada de Humahuaca" are of Spanish origin ($p.<0.05$).

The presence or absence of a Y-chromosome-specific Alu insert characterizes YAP+ and YAP- chromosomes, respectively (Hammer, 1994). The YAP+ insert, in association with other markers, is useful for identifying African or European ancestry (Hammer, 1995). All the cases of both samples were YAP-. Moreover, since none of the haplotypes referred to as undetermined in Table 1 have so far been observed in Y-chromosomes of African origin, it seems valid to conclude that none of the paternal lineages in our samples originated in Africa.

We determined the degree of genetic variability of Y-chromosomes according to Nei (1986,1987). Table 2 shows that the variability in "San Salvador de Jujuy" is significantly higher than in "Quebrada de Humahuaca" ($P < 0.001$).

Our results are in agreement with the ethno-historical data on human settlements in the Province of Jujuy as well as with the distribution of the GM phenotypes and haplotypes in the Jujeño populations located at different altitudes (Dipierri et al., 1999). The distribution of GM haplotypes corresponds to the 1778 census which showed that the Amerindian percentage was higher in Jujuy populations located at high altitude than in populations at lower levels. The opposite occurred with Negro, Mulatto and Spanish fractions.

Conclusions

The "San Salvador de Jujuy" population shows a significantly higher Y-chromosome variability than the one from "Quebrada de Humahuaca". These findings coincide with historical reports indicating that during the colonial period "San Salvador de Jujuy" was one of the main areas of gene admixture within the Province of Jujuy and Northwest Argentina.

CASES> 2500 M*	YAP	pSRY	αh	DYS19	DYS199	ORIGIN
16	-	C	II	A	T	Amerindian
1	-	C	1	B	T	Amenndian
1	-	C	II	Z	T	Amerindian
7	-	C	II	B	C	Caucasian
1	-	T	II	B	C	Caucasian
1	-	C	II	C	C	Caucasian
1	-	C	IV	C	C	Undetermined
CASES 1200 m*						
3	-	C	II	A	T	Amerindian
1	-	C	1	A	T	Amerindian
7	-	C	II	B	C	Caucasian
1	-	C	II	E	C	Caucasian
1	-	C	III	D	C	Undetermined
1	-	C	IV	B	C	Undetermined

* Figures are the total numbers of individuals having the same haplotype.

Table 1. Y-Chromosome Haplotypes.

Population	Total diversity (Ht)	Total intrapopulation diversity (Hs)	interpopulation diversity (Dst/Dsm) intrapopulation	Percentage of interpopulation diversity (Gst/Gst')	Subpopulation diversity (h)	Significance (Student Test)
	0.62	0.56	0.13	10.2		
> 2500 m					0.44± 0.034	
						P <0.001
1200 m					0.71±0.074	

* Estimates were according to Nei (1986, 1987)

Table 2. Y-Chromosome Genetic Diversity*.

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