

## **Beta-globin gene cluster haplotypes in the Mapuche Indians of Argentina**

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### **ABSTRACT**

Haplotypes derived from five polymorphic restriction sites in the beta-globin gene cluster were investigated in 86 chromosomes from the Argentinian Mapuche. These results were integrated with those previously obtained for ten Brazilian Indian tribes. Eight haplotypes were identified, the most frequent being 2 (57%) and 6 (27%). The presence of haplotype 3 in 2% of the Mapuche chromosomes is probably an evidence of admixture with individuals of African ancestry. Due to the high number of haplotypes observed, heterozygosity as measured by the Gini-Simpson index was higher in the Mapuche than in Brazilian Indians. The haplotypic distribution in the Mapuche was also significantly different from those of all Brazilian tribes investigated. This heterogeneity could be at least partially explained by admixture with non-Indian populations.

### **INTRODUCTION**

The beta-globin gene cluster was the first to be described and is still one of the most studied nuclear DNA segments used for

investigation of evolutionary relationships of human populations (Wainscoat *et al.*, 1986; Long *et al.*, 1990; Chen *et al.*, 1990). Among the aboriginal populations of the Americas, ten Brazilian Indian tribes were screened for this nuclear region (Guerreiro *et al.*, 1994; Bevilaqua *et al.*, 1995). Ten haplotypes were identified among these populations, but only two of them, 2 (+----) and 6 (-+--+), were frequent and present in all Brazilian tribes.

The Mapuche Indians are of special interest because they inhabit a large portion of southern Chile and Argentina, representing the southern geographical extreme of the Amerind migration. The present paper gives information about the beta globin gene cluster haplotypes in these Indians, and integrates these results with those already available for Amerindian populations.

## MATERIAL AND METHODS

The Argentinian Mapuche Indians are distributed along 211 localities, and number about 28,000 people (Carnese *et al.*, 1996). They are of Araucanian origin, and speak a language classified as Mapudugun (Carnese, 1995). Blood samples were collected in three communities. Anecon Grande is situated in the 25 de Mayo Department, Province of Rio Negro (69°30'W, 41°30'S), while the communities of Cerro Policia and Aguada Guzman (68°37'W, 39°10'S and 68°59'W, 30°39'S, respectively) are located in the El Cuy Department, in the north of the Province of Rio Negro. These communities all share the same habitat, rigorous winters in the Patagonian steppe.

A total of 43 subjects were investigated for five polymorphic restriction sites of the beta globin gene cluster: Hinc II<sub>5'</sub>ε, Hind III<sub>G</sub>γ, Hind III<sub>A</sub>γ, Hinc II<sub>φβ</sub> and Hinc II<sub>3'φβ</sub>. Blood samples were refrigerated shortly after collection and sent by air to Buenos Aires, where high molecular weight DNA was extracted using the salting out procedure described by Miller *et al.* (1988). PCR amplification conditions and primers used were as described in Bevilaqua *et al.* (1995).

The frequencies of polymorphic restriction sites and haplotypes were determined as described by Chen *et al.* (1990), and their distributions among tribes were compared by the Roff and Bentzen (1989)  $\chi^2$  test. Genetic diversity was measured by the Gini-Simpson index, which can be computed according to the formula:

$$GSI = 1 - \sum_k p_k^2$$

where  $p_k$  is the frequency of the  $k$ th haplotype in the sample (Rao, 1982; Long *et al.*, 1990). The index can be interpreted as the probability that two randomly sampled beta-globin haplotypes will differ in state.

## RESULTS AND DISCUSSION

Haplotype distributions obtained for the three Mapuche communities did not show significant differences (data not shown), and therefore they were grouped together. [Table 1](#) presents the haplotypes identified in all Amerindians investigated to date. Haplotypes 2 (+----) and 6 (-++++) were the most prevalent in the Mapuche and other populations. But among the Mapuche, haplotype 2 was somewhat less frequent (57%) than elsewhere, while haplotype 6 showed the highest frequency observed thus far in Amerindians (27%).

**Table 1 - Frequencies (%) of beta-globin gene cluster haplotypes observed in 11 South American Indian populations.**

Indian tribes (No. of chromosomes)	Haplotypes <sup>1</sup>										
	1 (-----)	2 (+----)	3 (----+)	4 (-+++)	5 (-++++)	6 (-++++)	7 (-++-)	9 (-++++)	11 (-+++)	15 (+++++)	16 (-+---)
Mapuche (86)	5.8	57.0	2.3	2.3	3.5	26.7	1.2	0.0	0.0	1.2	0.0
Xavante (60) <sup>2</sup>	0.0	60.0	0.0	0.0	15.0	18.3	5.0	0.0	0.0	0.0	1.7
Zoró (60) <sup>2</sup>	0.0	93.0	0.0	0.0	0.0	6.7	0.0	0.0	0.0	0.0	0.0
Gaviño (58) <sup>2</sup>	1.7	87.9	1.7	0.0	0.0	3.4	5.2	0.0	0.0	0.0	0.0
Sunú (44) <sup>2</sup>	0.0	81.8	0.0	0.0	4.5	11.4	0.0	0.0	2.3	0.0	0.0
Wai-Wai (56) <sup>2</sup>	0.0	87.5	0.0	0.0	0.0	12.5	0.0	0.0	0.0	0.0	0.0
Yanonami (34) <sup>2</sup>	2.9	91.2	0.0	0.0	0.0	5.9	0.0	0.0	0.0	0.0	0.0
Kayapo (44) <sup>2</sup>	0.0	79.5	0.0	0.0	0.0	20.5	0.0	0.0	0.0	0.0	0.0
Wayana Apaki (34) <sup>2</sup>	0.0	82.4	0.0	0.0	0.0	14.7	0.0	2.9	0.0	0.0	0.0
Wayampi (30) <sup>2</sup>	0.0	86.7	0.0	0.0	3.3	6.7	3.3	0.0	0.0	0.0	0.0
Araua (30) <sup>2</sup>	0.0	83.3	0.0	3.3	0.0	10.0	0.0	3.3	0.0	0.0	0.0

<sup>1</sup>Haplotype designation according to Long *et al.* (1990). <sup>2</sup>Data from Bevilacqua *et al.* (1995). <sup>3</sup>Data from Guerreiro *et al.* (1994).

Haplotypic diversity, as measured by the Gini-Simpson index (Long *et al.*, 1990), is shown in [Table II](#). The high value observed among the Mapuche was expected, because they have the highest number of haplotypes (8). Considering all South American tribes investigated thus far, 11 haplotypes have been identified ([Table I](#)). According to Long *et al.*'s (1990) classification, six of them (1-6) are first order or primitive, derived directly from the ancestral or another first order haplotype. The other five (7, 9, 11, 15 and 16) are considered as being of second order, having originated by recombination and/or gene conversion from first order haplotypes. On a global scale, they are less frequent in most populations in which they occur (Wainscoat *et al.*, 1986; Long *et al.*, 1990; Chen *et al.*, 1990).

Table II - Haplotypic diversity in South American Indians.		
Indian tribe	No. of haplotypes	GSI <sup>3</sup>
Mapuche	8	0.604
Xavante <sup>1</sup>	5	0.582
Kayapo <sup>2</sup>	2	0.325
Suruil	4	0.315
Wayana-Apala <sup>2</sup>	3	0.299
Arara <sup>2</sup>	4	0.293
Wayamp <sup>2</sup>	4	0.242
Gavião <sup>1</sup>	5	0.222
Wai-Wai <sup>1</sup>	2	0.218
Yanomami <sup>2</sup>	3	0.164
Zoró <sup>1</sup>	2	0.124
<sup>1</sup> Data from Bevilaqua <i>et al.</i> (1995). <sup>2</sup> GSI computed from data of Guerreiro <i>et al.</i> (1994). <sup>3</sup> GSI: Gini-Simpson index.		

A highly significant difference ( $\chi^2 = 54.18$ ;  $P = 0.0001$ ) was observed when the Mapuche were compared with the Brazilian Indians. Guerreiro *et al.* (1994) observed a homogeneous distribution among Amazonian tribes, while Bevilaqua *et al.* (1995) verified that the Xavante had a different haplotype distribution in relation to all others. The Mapuche also differ from the Xavante ( $\chi^2 = 17.13$ ;  $P = 0.013$ ) and from the other tribes ( $\chi^2 = 66.01$ ;  $P = 0.010$ ).

The presence of haplotype 3 in 2% of the Mapuche chromosomes suggests the occurrence of African-derived admixture among them, since this haplotype is absent from almost all other Indian groups studied, and it is relatively common in Africa. Evidence of non-Indian markers among the Mapuche was already obtained in other systems (Bailliet *et al.*, 1994; Hutz *et al.*, 1997; Bianchi *et al.*, 1997; Goicoechea, A.S., Carnese, F.R., Caratini, A.L., Avena, S., Salaberry, M. and Salzano, F.M., unpublished results).

How can these results be integrated with previous genetic information obtained in this Indian population? Some heterogeneity between northern and southern South American Indian populations was described at the protein and DNA levels (Haas *et al.*, 1985; Carnese *et al.*, 1996; Hutz *et al.*, 1997). The present investigation confirms these results; however, how much of this variation is due to non-Indian gene flow in the south or to real original differences can only be elucidated by further accumulation of genetic information on these groups.

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## RESUMO

Haplótipos derivados de cinco sítios de restrição polimórficos presentes no agrupamento da globina beta foram investigados em 86 cromossomos da população mapuche da Argentina. Esses resultados foram analisados em conjunto com os previamente obtidos para dez tribos indígenas brasileiras. Oito haplótipos foram identificados, dos quais os mais freqüentes foram o 2 (57%) e o 6 (27%). A presença do haplótipo 3 em 2% dos cromossomos dos Mapuches é uma evidência de mistura com indivíduos de ancestralidade africana. Devido ao alto número de haplótipos, a heterozigosidade medida pelo índice Gini-Simpson é mais alta nos Mapuches do que nos índios brasileiros. A distribuição haplotípica nos Mapuches é também significativamente diferente da observada nas tribos brasileiras. Essa heterogeneidade poderia ser parcialmente explicada pela mistura com populações não-indígenas.

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