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ANALYSES OF MITOCHONDRIAL DNA POLYMORPHISMS IN SKELETAL REMAINS AND EXTANT POPULATIONS OF NORTHERN CHILE

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Mitochondrial DNA polymorphisms have been extensively used in the reconstruction of the human evolutionary history owing to its maternal inheritance, lack of recombination, and high mutation rate relative to nuclear DNA. In the Americas, the primary goals of the mitochondrial DNA analysis have been to determine the origins, relationships and migrational patterns of New World populations. In this respect, the study of the frequency distribution of the four founding Amerindian haplogroups defined by Torroni et al. (1992), has proven useful. Each haplogroups can be characterized by a specific mtDNA marker: an Hae III restriction site gain at np. 663 for haplogroup A, the 9 bp. deletion in the COII/tRNAlys region for haplogroup B, a Hinc II restriction site loss at np. 13259 for haplogroup C and an Alu I restriction site loss at np. 5176 for haplogroup D. In order to expand the genetic data related to the peopling of South America, we studied mtDNA polymorphisms in five skeletal remains from the Azapa valley (AZ-71 site) dated 3000 to 2000 years BP, and compared them with the extant population of San Pedro de Atacama in northern Chile. The mtDNA of contemporary individuals was extracted by standard procedures (Lahiri DK et al. 1988). Ancient mtDNA was extracted from bone remains following an adapted protocol from Höss & Pääbo (1993). First, to destroy possible contaminant DNA on the surface of remain, bone fragments were exposed to UV irradiation. The samples were crushed and then powdered in a refrigerated mill. The powdered samples were incubated in 8 ml of 0.5 M EDTA solution for decalcification for 24 hours, and collected by centrifugation. The precipitate was suspended in 5 ml of lysis buffer (5.5 M guanidinium thiocyanate, 50 mM Tris-HCl pH: 6.4, 20 mM EDTA and 1.3 % Triton X-100) and incubated at 60ºC for several hours, with sporadic shaker. After centrifugation, the supernatant was removed to another tube and mixed with equal volume of lysis buffer. A 40 ml aliquot of silica suspension was added, and the mixture was incubated for 15 minutes at room temperature. After centrifugation the silica pellet was washed successively with washing buffer, 70% ethanol and acetone. Finally the pellet was dried, and the ancient DNA was eluted at 56ºC in 100 ml TE buffer.

The extant population of San Pedro de Atacama shows the four Amerindian haplotypes, analyzed by Amp-RFLP, with the following distribution: haplogroup A, 8.3%; haplogroup B, 62.5%; haplogroup C, 25% and haplogroup D, 4.2%. This distribution is very similar to the one described previously for Aymara populations from the North of Chile, and is in consistent with findings in other Andean high altitude populations. We identified, furthermore the sequence of the mtDNA hypervariable region I (position 16081 to 16409) for 23 individuals. Results revealed consistent changes described for these haplogroups.

We obtained a preliminary sequence analysis of hypervariable region I on mtDNA extracted from five mummies. These sequences showed a variable number of changes respect to the published sequence (Anderson et al. 1981). The five sequences present C-T transition at np. 16223, a characteristic change described in Amerindian haplogroups A, C and D, but absent in haplogroup B. Two of these sequences have transition T-C at np. 16298 and np. 16325. This change is characteristic of haplogroup C individuals. The other three individuals do not present all of the characteristic change that define the Amerindian haplogroups at the sequence level. Furthermore,

we found in all sequences the absence of the characteristic nucleotides changes defining haplogroup B. Based on the partial sequences obtained from these five mummies we may suggest that haplotype B frequencies in the ancient populations of northern Chile were lower then in Andean extant populations, of the same region. Extensive sequence analyses and Amp-RFLP must be performed in a larger sample in order to corroborate these results which are certainly to preliminary to draw microevolutionary conclusions.

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References Cited

Anderson, S., A.T. Bankier, B.G. Barrell, M.H.L. de Bruijn, A.R. Coulson, J. Drouin, I.C. Eperon, D.P. Nierlich, B.A. Roe, F. Sanger, P.H. Schreier, A.J.H. Smith, R. Staden, and I.G. Young 1981 Sequence and organization of the human mitochondrial genome. *Nature 290*: 457-465. [Links]

Höss, M., and S. Pääbo 1993 DNA extraction from pleistocene bones by a silica-based purification method. *Nucleic Acids Research* 16: 3913-3914. [Links]

Lahiri, D.K., and J.I. Nurnberger 1991 A rapid non-enzymatic method for the preparation of HMW DNA from blood for RFLP studies. *Nucleic Acids Research* 19: 5444. [Links]

Torroni, A., T,G, Schurr, C.C. Yang, E.J.E. Szathmary, R.C. Williams, M.S. Schanfield, G.A. Troup, W.C. Knowler, D.N. Lawrence, K.M. Weiss, and D.C. Wallace 1992 Native American mitochondrial DNA analysis indicated that the Amerind and the Nadene populations were founded by two independent migrations. *Genetics* 130: 153. [Links]

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