International Congress Series 1261 (2004) 625–627

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Uniparentally inherited genetic markers as tools for ethnic and geographical origin detection of forensic samples

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Abstract. Uniparentally inherited polymorphic genetic markers opened a new scope in the human forensic identification. These genetic markers may display geographic and/or ethnic specific characteristic features that may provide a clue about the origin of the donor of a given sample. Maternally inherited mitochondrial (mtDNA) polymorphisms and paternally transmitted Y-chromosome specific polymorphic sequences when investigated in combination may provide relevant information concerning the ethnic/geographical origin of a person. Some populations are particularly more suited for these investigations due to their differential attributes. South American aboriginal are a good example. In this contribution, we provide information about an isolated aboriginal community that depicts ethnic specific attributes. The combined use of matri and patri lineage markers allowed to estimate the admixture at the individual level.

Keywords: Ethnic group–DNA polymorphism; mtDNA; Y-chromosome polymorphisms

1. Introduction

Human identification by DNA typing is based on the analysis of highly polymorphic autosomal short tandem repeats (STRs). Although highly informative, these markers are unable to provide information concerning the ethnic or geographic origin of the donor of a sample. In those cases in which no suspects are available, such as rape cases, homicides or burglary, or when fragmentary human remains are the sole source of genetic material, as in suicide terrorist attacks cases, it would be desirable to detect at least the ethnicity or geographic origin of these evidentiary material. Uniparentally inherited genetic markers

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0531-5131/
doi:10.1016/S0531-5131(03)01524-3

(e.g. Y-chromosome specific or maternally inherited mitochondrial (mtDNA) polymorphisms) may provide a clue for such identification. Since males perpetrate most violent crimes, gender-specific genetic markers may represent a valuable identification tool [1]. Mammalian Y-chromosomes are characterized by the absence of recombination in the major extension of the chromosome (except PAR 1 and 2 in the telomers, of both arms) and by the presence of a wide range of different types of polymorphic loci. These markers include alphoid satellite variants, minisatellites (MYS1) and microsatellites (Y-STR) all of them with high mutation rate, as well as others, with low mutation rate, included within the so-called Unique Event Polymorphisms (UEP). These biallelic markers are the Single Nucleotide Polymorphism (Y-SNP) and the Insertion/deletion events (Ins/del), like Y Alu Polymorphism (YAP), and may provide additional information when analyzed in combination with highly variable Y-STRs. In order to investigate this relevant forensic aspect, different geographically isolated aboriginal communities inhabiting North Eastern Argentina were investigated. Three communities of the Guarani M'Byá tribe from different localities were investigated by Y-STR nonaplex (DYS19, DYS385, DYS389 I and II, DYS390, DYS391, DYS392 and DYS393), the ins/del YAP, the SNP DYS199, as well as the mtDNA Region V 9bp ins/del. In order to confirm the Amerindian mtDNA haplogroup B, HVRI sequencing of those samples with 9-bp deletion was carried out. The overall information was compared with the results obtained with a set of Argentinean samples randomly chosen from different regions of the country.

2. Materials and methods

2.1. Samples

Three communities belonging to the Guarani M'Byá tribe inhabiting Misiones Province (North Eastern Argentina) were selected, (M'Bororé, $N=52$; Yriapu, $N=10$; and Ybaté, $N=6$). Blood samples were obtained by finger puncture from unrelated male donors. In addition, 50 samples randomly chosen from different regions of Argentina were also analyzed.

Blood samples collected onto FTA paper were processed according with manufacturer's protocol.

Y-STRs: DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393 and DYS385 were amplified using one fluorescent dye labeled primer. Amplicons were detected with an ABI310.

DYS199: Amplification using allele-specific primers was performed according to Underhill (1996) [2]. The amplification products were separated in 2% agarose gels and detected by Ethidium Bromide staining through UV light.

Ins/del YAP: amplification using specific primers followed by agarose gel electrophoresis was employed.

mtDNA analysis: Hipervariable Region I was amplified, purified and then sequenced by means of Big Dye Terminator system, employing an ABI 310 sequencer.

Nine-base pair deletion mt DNA Region V was amplified as previously described [3] using forward primer labeled with TET. The detection was made in an ABI 310 sequencer.

3. Results and discussion

Within the sample investigated ($N = 68$ individuals) for the Y-STR minimal haplotype, a reduced value of haplotype diversity was detected (78%). Accordingly, a minimal Y-STR haplotype defined as: DYS19/13, DYS389I/14, DYS389II/31, DYS390/24, DYS391/11, DYS392/14, DYS393/11 and DYS385/14.16, denoted a high frequency (35%). This haplotype is neither present in other Argentina aboriginal tribes such as Mapuche, Tehuelche and Wichi nor in the Reference Y-STR Database http://www.ystr.charite.de) in which population data of Metropolitan areas of Argentina are also included [4]. This particular Guaraní haplotype is combined with the UEPs variants DYS199/T and YAP (−). When a reduced combined haplogroup including DYS19, DYS392, DYS393 and DYS199 is considered, a still higher frequency is detected (43%). This Ethnic Specific Combined Haplogroup (ESCH) may represent the effect of a bottleneck resulting in a founder effect that might be interpreted under the light of the history of this region. In addition, it may be used as an informative approach for tracing and identifying patrilineages from male-produced evidences, in forensic casework.

In contrast, the Metropolitan sample of Argentina shows a different picture. The haplotype diversity is, in this group, 98%, and the most frequent haplotype is represented in 3 out of 50 individuals (6%). Fourteen percent of these samples are DYS199/T in contrast with 87% in the Guaraní sample.

In average, 17% of the samples investigated in both groups depicted the mitochondrial Region V 9-bp deletion. The sequence of the HVR I confirmed that these samples belonged to the Amerindian B haplogroup [5]. Since in this work only haplogroup B was taken into account, further analysis would be required to assess the frequencies of the other Amerindian haplogroups.

The results emerging from the uniparentally inherited genetic markers might suggest: (a) that the Guarani community may represent a newcomer ethnic component, (b) that the metropolitan areas population has a much higher aboriginal component than expected and (c) the ESCH identified may represent a suitable male-specific haplogroup to be used in forensic casework.

Acknowledgements

This work was supported, in part, by grants 0746-CONICET, and UBACyT.B-038, to DC. DC and AS are members of CIC-CONICET.

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