



## Short communication

## Population genetic data of 38 insertion-deletion markers in six populations of the northern fringe of the Iberian Peninsula



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## ABSTRACT

Insertion-deletions have been reported very useful markers for forensic purposes. To further deepen in this matter, 38 non-coding bi-allelic autosomal indels were analyzed in 575 individuals representing six populations from the northern fringe of the Iberian Peninsula. Autochthonous populations from the Basque Country, northern Navarre, the Pas Valley in Cantabria and Aragon were analyzed, together with non-autochthonous populations from the Basque Country and northern Navarre. At the intra-population level, all loci analyzed were in Hardy-Weinberg equilibrium except for marker rs33917182 in autochthonous Basques. Linkage disequilibrium (LD) test did not reveal statistically significant allelic association between the different loci pairs in all six populations. Forensic parameters proved to be highly informative in the six populations analyzed, even if a scenario with population substructure and local inbreeding was considered for match probability calculations, and the potential of this indels set to be used in combination with other genetic markers is remarkable. As for inter-population analyses, in general terms the six populations showed low but statistically significant genetic distances. However, though this indels set efficiently differentiate between main ancestries, it does not allow an accurate separation at a local level and, for the time being, their combination with other informative markers is needed to maximize the power to accurately differentiate populations with close genetic ancestry.

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## 1. Introduction

The north of the Iberian Peninsula is home to a pool of singular populations characterized by striking archaeological, linguistic and genetic features within the European landscape. Some of these human groups, as the case of Basques, have been widely studied from a genetic perspective to find that the peculiarities of their genetic heritage might have some impact in forensic estimations. Mitochondrial DNA has shown a low diversity of maternal lineages in autochthonous populations from the Basque Country, Navarre or, especially, the Pas Valley [1–3]. STRs and SNPs from the Y-chromosome have also revealed distinctive patterns in some of these northern Iberian populations [4–7]. Additionally, their

genetic structure has been studied in detail for X-chromosome STRs [8,9] or autosomal markers [10]. However, insertion-deletion polymorphisms (indels) remain unexplored in these populations. These markers combine interesting characteristics of other genetic markers commonly used in forensics (STRs and SNPs). They are abundant and widely distributed along the human genome, have a lower mutation rate than STRs, can be analyzed in short amplicons and are easy to analyze by multiplex PCR and capillary electrophoresis [11], among other interesting features. All of this makes indels a promising type of genetic variation that may be of great interest as a complement to other genetic markers of forensic interest already in use. Furthermore, their contribution to obtain a higher power of discrimination could be even more significant in isolated populations with low gene diversities, such as autochthonous Basques or *Pasiegos* from the Pas Valley. Thus, the purpose of this study was to characterize the diversity of a set of 38 indel markers [11] in populations from the northern fringe of the Iberian Peninsula, and to assess their usefulness both in forensic casework and population genetics.

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## 2. Materials and methods

### 2.1. DNA samples

Autochthonous individuals from four populations of northern Iberian Peninsula were selected for analysis. The final pool included 100 Basques from the Basque Country, 102 Basques from Northern Navarre, 81 *Pasiegos* from the Pas Valley in Cantabria and 102 individuals from Aragon. Criteria for autochthony included local ancestry for at least three generations in the four populations and, additionally, a minimum of eight Basque surnames for both Basque populations. In addition to the autochthonous groups, we analyzed 100 individuals living in the Basque Country and 90 individuals residing in the Chartered Community of Navarre who did not meet the aforementioned criteria of autochthony since they had at least one ancestor from another region of the Iberian Peninsula. In all, we analyzed 575 individuals from the northern fringe of the Iberian Peninsula. As for Ethics Statement, all the samples were collected under informed consent, following procedures in accordance with the ethical standards of the Helsinki Declaration. The study was approved with the favorable ethical report from the Faculty of Pharmacy of the University of the Basque Country, signed at 26th September 2008. In addition, the study met with the approval of the Health Department of the Government of the Chartered Community of Navarre and the Scientific and Ethical Committee of the Basque Biobank for Research-OEHUN [www.biobancovasco.org](http://www.biobancovasco.org) that provided 74 of the 100 autochthonous Basques from the Basque Country.

Genomic DNA was obtained from saliva samples and peripheral blood samples using a salting out method. DNA quantitation was carried out using Quantifiler<sup>®</sup> Human DNA Quantification Kit (AB/LT/TFS: Applied Biosystems<sup>™</sup>, Life Technologies, ThermoFisher Scientific, Waltham, MA, USA).

### 2.2. Indel genotyping

The 38 indels were amplified in all the samples using the PCR protocol originally described by Pereira *et al.* [11] using a Veriti<sup>®</sup> Thermal Cycler (AB/LT/TFS). Fragment analysis of PCR products was

carried out in a 3500 Genetic Analyzer (AB/LT/TFS). Finally, GeneMapper<sup>®</sup> ID-X Software v1.2 (AB/LT/TFS) was used for allele designation.

### 2.3. Data analysis

Allele frequencies, heterozygosity ( $H$ ), Hardy-Weinberg equilibrium deviation, linkage disequilibrium and within-population inbreeding coefficient ( $F_{IS}$ ) were performed using Arlequin v3.5 [12]. As for statistical parameters of forensic interest, matching probability (MP) for each locus was calculated as  $P(A_iA_i, A_iA_i)$  and  $P(A_iA_j, A_iA_j)$  for homozygous and heterozygote genotypes, respectively, using formulae described in [13]. Power of discrimination (PD) was calculated as  $PD = 1 - MP$ . Power of exclusion (PE) for each locus was calculated using PowerStats v1.2 [14]. To examine the relationship of the populations under study and with data available for other populations [11,15,16], pairwise genetic distances ( $F_{st}$ ) were calculated and the resulting matrix was visualized in two multidimensional scaling (MDS) using PAST software v3.06 [17].

## 3. Results and discussion

A total of 1154 chromosomes were analyzed in this study. Genotypic data per sample and population are included as Supplementary material and statistic parameters for each population are shown in Supplementary Table 1. All loci analyzed in this study were in Hardy-Weinberg equilibrium after Bonferroni's correction for multiple tests ( $p > 0.00132$ ) with the exception of marker rs33917182 (B09) in autochthonous Basques. Linkage disequilibrium (LD) test did not show statistically significant allelic association between the different loci pairs in the same populations after Bonferroni's correction ( $p > 0.00132$ ). The lowest average value of expected heterozygosity was observed in Basque autochthonous (0.41441) and the Pas Valley (0.42255) (Table 1). Nonetheless, expected heterozygosity of the six populations was within the range of other populations analyzed for the same set of indels [11,18,19].

With regard to forensic parameters, the 38 indels set proved to be highly informative in the six populations analyzed (Table 1). If a

**Table 1**

Diversity and Forensic Parameters for the set of 38 indels analyzed in six populations from northern Spain. Expected values of forensic parameters under assumption of non random mating are shown ( $\theta = 0.01$  and  $\theta = 0.03$ , as recommended in the NRC II report, were used). In populations in which specific FIS index ( $f$ ) was closed to 0,  $f \approx 0$  was assumed.

	Obs Het	Exp Het	Combined MP	Combined PD	Combined PE
Basque autochthonous $\theta = 0.01, f \approx 0$ $\theta = 0.03, f \approx 0$	0.43553 ± 0.12269	0.41441 ± 0.08437	1.8951E-14 2.8138E-14 6.1906E-14	0.999999999999981 0.999999999999972 0.999999999999938	99.7625%
Basque residents $\theta = 0.01, f \approx 0$ $\theta = 0.03, f \approx 0$	0.43237 ± 0.08397	0.43629 ± 0.06543	3.9543E-15 5.8246E-15 1.2713E-14	0.999999999999996 0.999999999999994 0.999999999999987	99.7429%
Navarre autochthonous $\theta = 0.01, f = 1.50\%$ $\theta = 0.03, f = 1.50\%$	0.42999 ± 0.07832	0.43612 ± 0.06703	4.0314E-15 4.8702E-15 1.0783E-14	0.999999999999996 0.999999999999995 0.999999999999989	99.7082%
Navarre residents $\theta = 0.01, f \approx 0$ $\theta = 0.03, f \approx 0$	0.43840 ± 0.08941	0.43400 ± 0.07311	4.8914E-15 7.1690E-15 1.5503E-14	0.999999999999995 0.999999999999993 0.999999999999984	99.7970%
Pas Valley $\theta = 0.01, f = 1.44\%$ $\theta = 0.03, f = 1.44\%$	0.41650 ± 0.07461	0.42255 ± 0.07005	1.0238E-14 1.3081E-14 2.9737E-14	0.999999999999990 0.999999999999987 0.999999999999970	99.5379%
Aragon $\theta = 0.01, f \approx 0$ $\theta = 0.03, f \approx 0$	0.44157 ± 0.08171	0.43346 ± 0.06919	4.8534E-15 7.1496E-15 1.5596E-14	0.999999999999995 0.999999999999993 0.999999999999984	99.8045%

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