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Short communication

A genetic overview of Atlantic coastal populations from Europe and North-West Africa based on a 17 X-STR panel

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ABSTRACT

The forensic use of X-STRs requires the creation of allele and haplotype frequency databases in the populations where they are going to be used. Recently, an updated Spanish allele and haplotype frequency database for the new 17 X-STR panel has been created, being the only database available up to now for this new multiplex. In order to broaden the forensic applicability of the 17 X-STR panel, 513 individuals from four different populations located on the Atlantic Coast of Europe and North–West Africa have been studied, i.e. Brittany (France), Ireland, northern Portugal, and Casablanca (Morocco). Allele and haplotype frequency databases, as well as parameters of forensic interest for these populations are presented. The obtained results showed that the 17 X-STR panel constitutes a highly discriminative tool for forensic identification and kinship testing in the studied populations. Furthermore, we aimed to study if these populations located on the Atlantic coast actually share alike allele and haplotype frequency distributions since they have experienced genetic exchanges throughout history. This would allow creating larger forensic databases that include several genetically similar populations for its use in forensic casework. For this purpose, pairwise F_{ST} genetic distances between the analyzed populations and others from the Atlantic Coast previously studied with the 17 X-STR panel or the ten coincident markers included in the decaplex of the GHEP-ISFG were estimated. Our results suggest that certain nearby populations located on the European Atlantic coast could have underwent episodes of genetic interchange as they have not shown statistically significant differentiation between them. However, the population of Casablanca showed significant differentiation with the majority of the European populations. Likewise, the autochthonous Basque Country and Brittany populations have shown distinctive allele frequency distributions between them. Therefore, these findings seem to support that the use of independent allele and haplotype frequency databases for each population instead of a global database would be more appropriate for forensic purposes.

1. Introduction

The X-chromosomal short tandem repeats (X-STRs) have been widely studied over the last years by the forensic community [1]. These markers have been established as the perfect complement to the autosomal STRs (AS-STRs) and other lineage markers, such as Y-chromosomal STRs (Y-STRs) and mitochondrial DNA, when

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solving complex kinship cases [2]. Additionally, the X-STRs are of great utility in the identification of war victims and historical cases where the majority of the profiles to compare correspond to second or third degree relatives, such as grandparents-grandchildren, maternal uncles-nephews, etc. [3].

The forensic application of X-STRs requires the creation of allele and haplotype frequency databases in the populations where they are going to be used. Since the establishment of the decaplex of the GHEP-ISFG [4,5] and the Investigator™ Argus X-12 Kit (Qiagen GmbH, Hilden, Germany), many populations have been studied using these two multiplexes [1]. Recently, a new 17 X-STR panel in a single PCR reaction has been developed and validated [6] and, up to now, an updated Spanish allele and haplotype frequency database for this panel has been published [7]. However, the application of the new multiplex in other populations requires the creation of new databases.

The objective of this work was to broaden the forensic applicability of the 17 X-STR panel to other populations. For this purpose, allele and haplotype frequency distributions of four populations located on the Atlantic Coast of Europe and North-West (NW) Africa have been studied in order to enlarge the current X-STR databases for their application in forensic casework.

2. Materials and methods

2.1. Population samples

A total of 513 unrelated individuals (500 men and 13 women) were selected for this study. This sample set comprised individuals from four different regional populations located on the Atlantic Coast of Europe and NW Africa, i.e. Brittany (N = 179; XY = 179), Ireland (N = 100; XY = 100), northern Portugal (N = 79; XY = 79), and Casablanca (N = 155; XX = 13; XY = 142) (Supplementary Fig. S1). Samples from Brittany were provided by the Université de Bretagne Occidentale (Brest, France), samples from Ireland by the National Neuroscience Centre (Beaumont Hospital, Dublin, Ireland), samples from northern Portugal by the National Institute of Legal Medicine and Forensic Sciences (Porto, Portugal), and samples from Casablanca by the University of Hassan II (Casablanca, Morocco). All the samples were obtained from volunteer donors under informed consent, following the ethical standards of Helsinki Declaration.

2.2. PCR amplification, electrophoresis and data analysis

The 17 X-STR markers (DXS8378, DXS9898, DXS7133, GATA31E08, GATA172D05, DXS6801, DXS7423, DXS6809, DXS6799, DXS7132, DXS9902, DXS6800, DXS6789, DXS10075, DXS10079, DXS6807, and DXS6803) were amplified in a single PCR reaction as described in [6]. Amplification of samples and performance of the PCR reaction were verified by conventional agarose gel electrophoresis. Capillary electrophoresis of PCR products was conducted on an ABI PRISM 3130 Genetic Analyzer with the POP7 polymer (Thermofisher Scientific, Waltham, MA, USA). Electrophoresis data were analyzed with GeneMapper ID software version 4.0 (Thermofisher Scientific, Waltham, MA, USA).

2.3. Confirmation of new allele variants

Previously not detected allele variants were checked by size comparison with other samples that presented neighboring alleles. With this purpose, genomic DNA of the compared samples was blended, amplified together in a single PCR reaction and checked by capillary electrophoresis.

2.4. Forensic parameters and statistical analysis

Allele frequencies and gene diversity (GD) were calculated for the four different populations analyzed herein from both male and female samples, when applicable. On the other hand, male samples were used to calculate haplotype frequencies for the cluster DXS7132-DXS10075-DXS10079 (as established in a previous study for the 17 X-STR panel [7]) and to perform the pairwise linkage disequilibrium (LD) tests. All aforementioned parameters were estimated by using the Arlequin software v.3.5.1.2 [8]. In addition, paternity exclusion index in duos (MEC_D) and trios (MEC_T) [9], as well as power of discrimination in males (PD_M) and females (PD_F) were calculated for the analyzed populations by using the online tool of the Forensic ChrX Research database (http://www.chrx-str.org).

2.5. Population comparisons

Pairwise F_{ST} genetic distances between the four populations studied and other populations previously studied with the 17 X-STR panel located on the Atlantic Coast from northern Iberian Peninsula were estimated. For this comparison, genetic profiles of Galicia and the Basque Country were taken into account [6,7] (Supplementary Fig. 1).

Samples from the Basque Country population were classified in two groups according to previous studies that have detected intrapopulation differentiation [10–14]: 1) autochthonous Basque population, that includes individuals with ancestor's surnames that support maternal and paternal Basque ancestry for at least three generations; and 2) resident Basque population, that correspond to individuals living in the Basque Country. The criteria for this classification were based on the information collected from each donor. The genetic distance between these two groups was calculated to analyze if there are enough significant differences between them to consider them as independent.

Additionally, the pairwise F_{ST} genetic distances among the above-mentioned populations and six more collected from the bibliography, typed with the decaplex of the GHEP-ISFG [4,5], were calculated. The following populations were selected by its nearby location to the Atlantic Coast in the Iberian Peninsula: northern and central regions of Portugal [5], Asturias [15], Cantabria [5], Pas Valley (Cantabria) [16], and autochthonous Basques from Navarre [17]. For this calculation, only the coincident markers between the

Table 1
Average gene diversity (GD) and the following combined values of forensic parameters for each population: combined power of discrimination in males (cPD_M) and females (cPD_F), and combined mean exclusion chance in father/daughter duos ($cMEC_D$) and trios involving daughters ($cMEC_T$). Studied populations: Brittany (BRIT), Ireland (IRE), northern Portugal (PORT), and Casablanca (MOR). N = number of X chromosomes.

	BRIT (N = 179)	IRE (N = 100)	PORT (N = 79)	MOR (N = 168)
GD	0.7313 ± 0.0565	0.7377 ± 0.0657	0.7301 ± 0.0701	0.7453 ± 0.0757
cPD_F	0.99999999999999992	0.99999999999999993	0.9999999999999998	0.999999999999999991
cPD_M	0.9999999997	0.9999999998	0.9999999995	0.99999999991
$cMEC_T$	0.999999998	0.999999998	0.999999996	0.9999999994
$cMEC_D$	0.9999993	0.9999994	0.9999990	0.9999998