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Analysis of six tetranucleotide polymorphisms of the X-chromosome in different Spanish regions

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Abstract. We studied 6 X-linked microsatellites in a large group of individuals from five Spanish regions. Those tetranucleotide markers (DXS9895, DXS9898, DXS7130, DXS7131, GATA172D05, and DXS6789) were amplified in two triplex PCR. There was no evidence for statistically significant linkage disequilibrium. There were no significant sex- or region-related differences in allelic frequencies, suggesting that general national databases can be adequate as a reference in X-linked markers. © 2005 Published by Elsevier B.V.

Keywords: STR; Microsatellites; X-chromosome; Haplotypes

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1. Introduction

Microsatellites of the X-chromosome are being increasingly studied in recent years as a useful tool in forensic analysis. Since fathers transmit the same X-chromosome to all their daughters, they are particularly useful in deficiency paternity cases when the child is a female, in maternity testing, and in paternity cases involving blood relatives [1]. Although frequencies from different continents have been published, it is unclear to which extent X-

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linked markers display differences in the mid-geographical range. Therefore, we decided to study the allelic frequency distribution in several populations distributed from the North part through the South part of Spain.

2. Materials and methods

We studied 614 subjects living in 5 Spanish regions (Cantabria, Basque Country, Galicia, Castilla and Andalucia).

Microsatellites were amplified in two triplex reactions: one for markers DXS7130, DXS7132 and DXS6789; another for markers DXS9895, DXS9898 and GATA172D05. The PCR products were injected into an ABI prism 310 capillary electrophoresis system (Applied Biosystems). Some common alleles were sequenced as previously reported to help in allele typing [2].

Sex and population differences in allelic frequencies were tested by a Monte Carlo extension of the Fisher exact test, using SPSS software. Disequilibrium linkage and Wright's Fst values for population stratification were estimated with GDA software. The polymorphism information content (PIC) was estimated as $1-\sum Pi^2-\sum\sum Pi^2Pj^2$. The average power of discrimination (PD) and the expected probability of exclusion (PE) were estimated with the following formulas proposed by Desmarais et al. [3].

3. Results

There were no sex-related differences in allelic frequencies. Likewise, the frequency distributions were very similar in all the populations studied. The overall Fst value was 0.009.

Data about the forensic efficiency of genetic markers are shown in Table 1. We did not find definitive evidence for linkage disequilibrium among the loci studied.

4. Discussion

The 6 X-linked markers reported in this paper can be amplified in two PCRs and may be quite useful in forensic cases. As it is the case with autosomic markers, these X-linked loci did not reveal a significant population stratification. A note of caution is usually raised about the lack of independence when interpreting the results of several markers located on the same chromosome. Indeed, some linkage disequilibrium among several X-linked markers has been previously reported [4]. However, despite theoretically possible, in the present study we did not find statistical evidence for such a linkage. The X-chromosome

Table 1
Forensic efficiency of the markers studied

	DXS7130	DXS7132	DXS6789	DXS9895	GATA172D05	DXS9898
PIC	0.716	0.739	0.816	0.727	0.790	0.753
PD female	0.894	0.903	0.904	0.894	0.934	0.911
PD male	0.733	0.758	0.752	0.748	0.805	0.772
PE trio	0.699	0.719	0.718	0.705	0.777	0.735
PE motherless	0.560	0.583	0.582	0.566	0.653	0.601

PIC, polymorphism information content; PD, power of discrimination; PE, power of exclusion.

appears to be rather prone to recombination, and an absence of linkage has been reported in other studies, even when quite close markers were studied [5].

In conclusion, this set of tetranucleotide X-linked STRs appears to be quite useful in forensic cases: they do not show clear evidence for disequilibrium linkage; they are quite discriminating; and general databases seem to be adequate for reference.

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