Int J Legal Med (2013) 127:63–64
DOI 10.1007/s00414-012-0672-z

POPULATION DATA

Population data of 12 X-STR loci in a North of Portugal sample

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Received: 5 December 2011 / Accepted: 13 January 2012 / Published online: 2 February 2012

DNA markers on the X-chromosome (ChrX) have been shown to be powerful tools for solving complex relationship cases [1].

The ChrX contains four linkage groups located at Xp22.2, Xq12, Xq26, and Xq28 that can provide independent genotype information. At present, it is proposed that it is preferable to use clusters to define haplotypes in forensic practice [2].

The X-Decaplex (DXS8378, DXS9902, DXS7132, DXS9898, DXS6809, DXS6789, DXS7133, GATA172D05, GATA31E08, and DXS7423) has been used by the GHEP-ISFG to make several studies [3, 4] and to solve forensic cases.

The Investigator Argus X-12 kit (Qiagen) includes 12 X-STRs located in four different linkage groups (linkage group 1: DXS10148, DXS10135, DXS8378; linkage group 2: DXS7132, DXS10079, DXS10074; linkage group 3: DXS10103, HPRTB, DXS10101; linkage group 4: DXS10146, DXS10134, DXS7423).

The Investigator ArgusX-12 Kit is a multiplex application kit that amplifies 12X-chromosomal STR loci highly suited for paternity testing [5], forensic applications, as well as population genetics and anthropological studies [6]. The analysis of complex kinship cases often requires the study of more genetic markers than the commonly used autosomal short tandem repeat systems to unravel simple cases [7].

DNA was extracted from buccal cell swabs by Chelex method [8] from 150 healthy unrelated males and 73 females from the North of Portugal, involved in paternity testing, after consent. The extracted DNA was amplified using the Investigator Argus X-12 kit (Qiagen), according to the manufacturer's recommendations (Investigator Argus X-12 Handbook, Qiagen®, 2010), in a GeneAmp 9700 PCR system (Applied Biosystems, Foster City, CA). Control DNA XX28 (Qiagen) was genotyped as standard reference. The amplified products were detected and separated by capillary electrophoresis in an ABI PRISM 3100 Genetic Analyzer (Applied Biosystems). Fragment sizes and genotypes were determined automatically using the GeneMapperID (Applied Biosystems). Allele designations were determined by comparison of the amplified fragments with those of the allelic ladders.

Allele frequencies, homozygotie (h), heterozygosity (HET), power of exclusion (PE), paternity index (PI), polymorphism information content (PIC), mean exclusion chance (MEC), and power of discrimination (PD) calculations were performed using http://www.chrx-str.org/. Our laboratory participates in the collaborative quality control and proficiency testing exercises of the GEP-ISFG group (http://www.isfg.org).

Here, we report allele frequencies and haplotype data of a North of Portugal population, involving 150 males and 73 females and 150 males, respectively.

The allele frequencies for the 12 X-STRs loci are shown in Table S1 and population statistic parameters in Table S2.

Haplotype frequencies were counted for each linkage group. The distribution of STR haplotypes are summarized in Table S3 and detailed haplotype data is supplied as supplementary material (Tables S4a, b, c and d).

Testing a population of 223 unrelated individuals (150 males and 73 females) revealed 129 different haplotypes for linkage group 1, 92 for linkage group 2, 86 for linkage group 3, and 116 for linkage group 4. None of them exceeds a frequency of 0.04, and the frequency of the unique haplotypes

Electronic supplementary material The online version of this article (doi:10.1007/s00414-012-0672-z) contains supplementary material, which is available to authorized users.

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for all the linkage groups is 0.00667. The unique haplotypes correspond to 85.3%, 64.1%, 67.4%, and 76.0% for the linkage groups 1–4, respectively.

The results support that ArgusX-12 Kit is very informative for forensic purposes. It can considerably improve the statistical information in forensic routine analysis, mainly because using linkage groups with three loci increases the power of discrimination compared to other available commercial kits. Comparing the Argus X-12 kit with the previously mentioned X-Decaplex [4], the matching probability in male samples, considering the loci as independent, decreased from 1.03×10^{-6} to 5.87×10^{-10}, and the power of discrimination increased from 99.9999% to 99.9999999%.

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