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Forensic validation of Y-chromosome STR polymorphisms in Italy: the GE.F.I. collaborative database

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Abstract

Haplotype data of 1176 Italian males from 10 regions were obtained as a part of a collaborative validation exercise. Individual data are available at http://www.gefi-forensicDNA.it.

Keywords: Forensic validation; Y-chromosome; STR polymorphism

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1. Introduction

The Italian group of forensic geneticists (GE.F.I.) organized a collaborative study to validate the use of Y-chromosome haplotypes in forensic practice. Laboratories participating to the exercise were asked to type at least 50 unrelated subjects born in their region for eight predefined STR loci, namely DYS19, DYS389-I, DYS389-II, DYS390, DYS391, DYS392, DYS393, and DYS385 [1,2]. The organizing committee sent four blind control samples to each group. Laboratories were left free to use their preferred experimental procedures.

2. Quality control

The following five samples were sent to each participating laboratory: (1) male DNA reference sample, with genotype results; (2) male stain (single profile); (3) Male stain (single profile); (4) admixture male–male; (5) admixture male–female. Thirteen laboratories sent results. The two single-profile blind controls were correctly identified in all cases, though in three instances, the marker DYS392 required a second amplification trial. Even the male–female admixture was easily typed (only one error in a lab, for marker DYS385); on the other hand, the male–male admixture was generally problematic, causing several interpretation errors, mainly for markers DYS392 and DYS385. It is worth noting that DYS392 is the only trinucleotide repeat locus of the series, and DYS385 is a composite locus. Data from these 13 laboratories were included in the database.

3. Results

Haplotype data from different laboratories of the same region were pooled. The final database included a total of 1176 male profiles from 10 Italian regions, embracing 68.2% of the Italian population [3]. The total number of different haplotypes was 960 (82%), and the number of unique haplotypes was 862 (73%). The frequency of the most frequent haplotype in the entire sample (n=30) was 0.026. This haplotype was the most frequent in 8 out of 10 regions, and is also the more frequent in Iberian and Center-European populations (YHRD Europe on-line database).

4. Conclusions

A low degree of heterogeneity is present among Italian regions for the frequencies of Y-chromosome haplotypes. A validated database of Y-chromosome haplotypes is now available for use in Court cases and in whatever forensic instance.

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