Announcement of population data

Regional Italian Allele frequencies at nine short tandem repeat loci

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Received 13 October 1999; received in revised form 15 May 2000; accepted 16 May 2000

Abstract

An Italian population study was performed on the loci D3S1358, vWA, FGA, TH01, TPOX, CSF1PO, D5S818, D13S317, D7S820 and a portion of the X-Y homologous gene Amelogenin for gender determination using the AmpFlSTR Profiler kit (PE Biosystems, Foster city, CA). This study was done on a population of 618 unrelated Italian individuals from 18 regions in Italy (except for Valle d’Aosta and Sardinia) to determine allele frequencies for each STR locus, and to evaluate STR technology for developing an Italian Offender DNA database.

Keywords: DNA; STR; Italian population; AmpFlSTR profiler; Forensic identification

1. Population samples

All individuals were examined and their parents were born in the same region. Whole blood samples were collected, with the cooperation of the Centro Analisi e Ricerche della Direzione Centrale di Sanità della Polizia di Stato from at least eleven individuals from each region.

2. DNA extraction and quantification

Chelex extraction procedure [1]. The extracted DNA was quantitated using the QuantiBlot kit (PE Biosystems) and chemiluminescent detection with ECL (Amersham).

3. PCR amplification

1–2 ng of target DNA following the protocols described in the AmpFlSTR Profiler User’s Manual (PE Biosystems). The samples were amplified using GeneAmp PCR System 9600 and GeneAmp PCR System 2400 (PE Biosystems) to compare the two thermal cyclers.
4. Electrophoresis and detection

ABI Prism 377 DNA Sequencer. Gel (36 cm wide, 0.2 mm thick) was 5% Long Ranger (FMC BioProducts, Rockland, ME) containing 6M urea and 1X TBE. Electrophoresis was for 2.5 h at 3000 V.

5. Typing

Sample analysis were accomplished using GeneScan Analysis software v.2.1, and genotypes were determined by comparison to allelic ladder alleles run on the same gel using Genotyper v.2.0.

6. Results

The results are shown in Table 1.

7. Statistical analysis

Statistical tests were performed to test for Hardy–Weinberg equilibrium (HWE) and independence between the loci by FBI software [2–9]. The probability of identity across all nine loci in this Italian population was estimated as 1 in 4.5 billion [10] (see Table 1).

<table>
<thead>
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<th>Allele</th>
<th>D3S1358</th>
<th>vWA</th>
<th>FGA</th>
<th>TH01</th>
<th>TPOX</th>
<th>CSF1PO</th>
<th>D5S818</th>
<th>D13S317</th>
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</table>

Probability of identity (PI)

\[ PI_{tot} = \frac{1}{4.5 \times 10^9} \]
8. Other remarks

Off-ladder variants were observed at the FGA locus (alleles 21.2, 22.2, 23.2 and 24.2) and the TH01 locus (allele 11). Regional differences were observed at the TH01 locus (see Table 2). There is a decline from North to South confirmed by test of homogeneity (RXC contingency table test with 2000 shuffling experiments — data not shown) and the North is significantly different from the South ($p < 10^{-3}$), and North compared to Sicily is different ($p < 0.005$). The Italian TH01 allele frequencies reported by Garofano et al. [11] are most similar to the North region data reported here. No detectable association between loci was observed, thus providing support for reliable use of the product rule to estimate multilocus profile frequencies. Those frequencies can be used in the database in the combined DNA Index System (CODIS by FBI) which is installed at the Forensic Science Service, Italian National Police, Ministry of the Interior.

Acknowledgements

The author would like to thank Gabriele Mauro for sampling of blood samples and the members of staff of the extraction, quantification and amplification areas. This work was supported by grants from Ministry of the Interior, Dipartimento della Pubblica Sicurezza.

References


Table 2

Observed Allele frequency for the locus TH01 in different area

<table>
<thead>
<tr>
<th>Allele</th>
<th>North</th>
<th>Central</th>
<th>South</th>
<th>Sicily</th>
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<td>5</td>
<td>0.394$^a$</td>
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<td>–</td>
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<td>33.990$^a$</td>
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<td>14.211$^a$</td>
<td>15.517$^a$</td>
<td>15.816$^a$</td>
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<td>8</td>
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<td>12.105$^a$</td>
<td>11.330$^a$</td>
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<td>16.837$^a$</td>
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<td>3.061$^a$</td>
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<td>0.246$^a$</td>
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</table>

Observed homozygosity 17.3% 14.7% 20.7% 19.4%

Experimental homozygosity$^b$ 21.3% 21.3% 21.6% 19.6%

Homozygosity test$^c$ 0.270 0.027 0.757 0.957

Likelihood test$^c$ 0.752 0.076 0.367 0.955

Exact test$^c$ 0.787 0.104 0.344 0.962

$^a$ These values are in percentage.

$^b$ Unbiased estimate of the expected homozygosity.

$^c$ Probability values.
