International Congress Series 1288 (2006) 283–285

www.ics-elsevier.com

# Population study of four X-chromosomal STR loci in the UK and Irish population

C. Oguzturun, C.R. Thacker *, D. Syndercombe Court

*Centre for Haematology, ICMS, Barts and The London, Queen Mary's School of Medicine and Dentistry, 4 Newark Street, London, E1 2AT, United Kingdom*

**Abstract.** X-chromosome STR typing can complement existing DNA profiling protocols and can also offer useful information in cases of complex kinship analysis. In this study a database relevant to the UK population was compiled. The four X-chromosome short tandem repeats DX8378, DX7132, DX7423 and HPRTB were used to generate allele frequencies in a population sample of 600 unrelated males and females from the UK and Irish populations. The population was composed of three subsets of data: 200 individuals who described themselves as Irish Caucasian; 200 individuals who described themselves as British Caucasian and 200 individuals who described themselves as South Asian (originating mainly from the countries of Bangladesh and Pakistan). Amplification was performed using the Mentype® Argus X-UL PCR amplification kit (Biotype AG). Slight modifications were made to the manufacturer's recommended protocol. Products were detected using ABI PRISM® 3100 Genetic Analyzer (Applied Biosystems). Allele frequencies were calculated and the three population subsets compared. The Mentype® Argus X-UL PCR amplification kit was found to be a robust system and a useful addition to autosomal markers routinely used in forensic and paternity testing applications. © 2006 Published by Elsevier B.V.

*Keywords:* X-chromosome STR; Population data; Mentype® Argus X-UL PCR amplification kit

## 1. Introduction

The analysis of short tandem repeats (STRs) in the field of human identification is now routine practice within the majority of forensic and paternity testing laboratories. Complex relationships, previously difficult to resolve, have benefited from the increase in experience and understanding of autosomal and Y-chromosome STR markers but restrictions still exist.

* Corresponding author. Tel.: +44 20 7882 2282; fax: +44 20 7882 2183.
*E-mail address:* c.r.thacker@qmul.ac.uk (C.R. Thacker).

0531-5131/ © 2006 Published by Elsevier B.V.
doi:10.1016/j.ics.2005.09.175

X-chromosomal STRs can go some way to minimise these limitations, particularly in deficient paternity cases involving a female child. The ability of gonosomal markers to assist in such cases, combined with the added expectation placed upon scientists to resolve complex issues of kinship, makes an increase in demand for the use of X-chromosome markers inevitable. Relevant population data is essential if accurate and appropriate conclusions are to be drawn regarding identification of samples and relationships between individuals. It was for this reason that the three datasets used in this study were selected. The Mentype® Argus X-UL PCR amplification kit (Biotype AG) allows simultaneous amplification of the four tetranucleotide X-chromosome STRs DX8378 (Linkage Group 1), DX7132 (Linkage Group 2), HPRTB (Linkage Group 3) and DX7423 (Linkage Group 4). Primers are labelled with 6-Fam making simultaneous detection with other markers a possibility.

## 2. Materials and methods

### *2.1. Samples and DNA extraction*

Buccal swabs and blood samples were collected from 600 unrelated, consenting individuals and three databases compiled. Database one was composed of samples donated by individuals originating from, resident in and describing themselves as Irish Caucasian. Database two was also comprised of Caucasian individuals with the population drawn from individuals originating from and resident in mainland Britain. Samples identified as South Asian in origin comprised database three. These samples were donated from unrelated individuals resident in mainland Britain originating mainly from Bangladesh and Pakistan. Identification and categorisation was made on the basis of personal classification, official documentation and provided photographs. DNA was extracted using Chelex®100 chelating resin. Quantification was not performed.

### *2.2. DNA amplification, detection and analysis*

Amplification was performed using the Mentype® Argus X-UL PCR kit (Biotype AG). The extracted DNA (1.2 μL) was amplified, in the presence of Platinum® Taq DNA Polymerase (Invitrogen), in a total PCR reaction volume of 10 μL. The GeneAmp® PCR System 9700 (Applied Biosystems) was set at a ramping speed of 1 °C/s (9600 emulation mode) and a total of 29 cycles performed. Products were detected using the ABI PRISM® 3100 Genetic Analyzer (Applied Biosystems) and the denaturing polymer POP-6. The supplied ROX 550 internal size standard was used and the provided sequenced ladder utilised for genotype classification. Positive (control DNA XY1 and XX74) and negative controls were run with each set of amplifications. For each dataset, allele frequencies were calculated and comparisons made using the exact test. Male and female genotypes were separated prior to calculations being performed.

## 3. Results and discussion

Generated allele frequencies are shown in Table 1. Intermediates and off ladder alleles were observed in all studied populations and at all loci except DX7423. No difference was observed between males and females ($p > 0.05$). No significant differences were seen between the two Caucasian populations, but they differed from the South Asian populations at DX8378 and DX7423 ($p < 0.001$).

Table 1
Allele frequencies for four X-chromosomal STR markers in three datasets relevant to the UK population

| Allele | British Caucasian | | | Irish Caucasian | | | South Asian | | |
|---|---|---|---|---|---|---|---|---|---|
| | M (150) | F (184) | P (289) | M (106) | F (182) | P (288) | M (140) | F (108) | P (248) |
| *DX8378* | | | | | | | | | |
| 8 | – | – | – | 0.0094 | – | 0.0035 | 0.0071 | – | 0.0040 |
| 9 | 0.0095 | 0.0217 | 0.0173 | – | 0.0165 | 0.0104 | 0.0500 | 0.0093 | 0.0323 |
| 9.3 | 0.0095 | – | 0.0035 | – | – | – | – | – | – |
| 10 | 0.2857 | 0.3533 | 0.3287 | 0.4528 | 0.3571 | 0.3924 | 0.2929 | 0.2222 | 0.2621 |
| 11 | 0.3524 | 0.3152 | 0.3287 | 0.3491 | 0.3022 | 0.3194 | 0.3429 | 0.4444 | 0.3871 |
| 12 | 0.3238 | 0.2609 | 0.2837 | 0.1698 | 0.3077 | 0.2569 | 0.2571 | 0.3056 | 0.2782 |
| 13 | 0.0190 | 0.0380 | 0.0311 | 0.0189 | 0.0165 | 0.0174 | 0.0500 | 0.0185 | 0.0363 |
| 14 | – | 0.0109 | 0.0069 | – | – | – | – | – | – |
| *HPRTB* | | | | | | | | | |
| 181.9 | – | 0.0054 | 0.0035 | – | – | – | – | – | – |
| 10 | 0.0286 | 0.0163 | 0.0208 | – | 0.0055 | 0.0035 | – | – | – |
| 10.2 | – | – | – | 0.0094 | 0.0110 | 0.0104 | – | – | – |
| 11 | 0.1524 | 0.1304 | 0.1384 | 0.0849 | 0.1044 | 0.0972 | 0.0857 | 0.1019 | 0.0927 |
| 11.2 | – | – | – | – | – | – | 0.0214 | – | 0.0121 |
| 12 | 0.2952 | 0.3152 | 0.3080 | 0.3774 | 0.3132 | 0.3368 | 0.3214 | 0.2963 | 0.3105 |
| 13 | 0.3524 | 0.3207 | 0.3322 | 0.2358 | 0.3462 | 0.3056 | 0.3571 | 0.3889 | 0.3710 |
| 14 | 0.1619 | 0.1467 | 0.1522 | 0.2075 | 0.1648 | 0.1806 | 0.1429 | 0.1389 | 0.1411 |
| 15 | 0.0095 | 0.0543 | 0.0381 | 0.0566 | 0.0440 | 0.0486 | 0.0714 | 0.0648 | 0.0685 |
| 16 | – | 0.0109 | 0.0069 | 0.0283 | 0.0110 | 0.0174 | – | 0.0093 | 0.0040 |
| *DX7423* | | | | | | | | | |
| 12 | – | – | – | – | 0.0110 | 0.0069 | – | – | – |
| 13 | 0.0952 | 0.0761 | 0.0830 | 0.1415 | 0.1319 | 0.1354 | 0.0071 | 0.0463 | 0.0242 |
| 14 | 0.3238 | 0.3315 | 0.3287 | 0.3302 | 0.3352 | 0.3333 | 0.4429 | 0.4352 | 0.4395 |
| 15 | 0.3810 | 0.4022 | 0.3945 | 0.3396 | 0.3846 | 0.3681 | 0.4143 | 0.4352 | 0.4234 |
| 16 | 0.1619 | 0.1359 | 0.1453 | 0.1509 | 0.1209 | 0.1319 | 0.1143 | 0.0648 | 0.0927 |
| 17 | 0.0381 | 0.0543 | 0.0484 | 0.0377 | 0.0110 | 0.0208 | 0.0143 | 0.0093 | 0.0121 |
| 18 | – | – | – | – | 0.0055 | 0.0035 | 0.0071 | 0.0093 | 0.0081 |
| *DX7132* | | | | | | | | | |
| 11 | – | 0.0109 | 0.0069 | – | 0.0110 | 0.0069 | 0.0143 | 0.0278 | 0.0202 |
| 12 | 0.0952 | 0.1033 | 0.1003 | 0.0849 | 0.1044 | 0.0972 | 0.1643 | 0.1019 | 0.1371 |
| 13 | 0.2571 | 0.3152 | 0.2941 | 0.3113 | 0.2637 | 0.2813 | 0.2571 | 0.1574 | 0.2137 |
| 13.3 | 0.0095 | – | 0.0035 | – | – | – | – | – | – |
| 14 | 0.3524 | 0.4022 | 0.3841 | 0.3113 | 0.3462 | 0.3333 | 0.3571 | 0.3889 | 0.3710 |
| 15 | 0.2571 | 0.1304 | 0.1765 | 0.2453 | 0.1978 | 0.2153 | 0.1643 | 0.2500 | 0.2016 |
| 16 | 0.0286 | 0.0326 | 0.0311 | 0.0377 | 0.0714 | 0.0590 | 0.0357 | 0.0556 | 0.0444 |
| 17 | – | 0.0054 | 0.0035 | 0.0094 | 0.0055 | 0.0069 | 0.0071 | 0.0185 | 0.0121 |

M, males; F, females; P, pooled (number of chromosomes tested).