



Haplotype discrimination amongst three UK population groups using three multiplexes to type eleven Y chromosome STRs

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Abstract

In certain circumstances, analysis with Y chromosome markers provides potential benefits over autosomal STRs, for example, paternity cases in which the putative father is deceased and in rape cases. The polymorphism level of Y chromosome STRs, however, is generally quite low and discrimination is further reduced as a result of linkage. Consequently, profiles need to be analysed as haplotypes rather than independent loci. To improve the current level of discrimination, we extended the range of Y-chromosome STRs most commonly used in forensic analysis, with the addition of three recently reported STR markers: DYS437, 438, and 439 to give a total of 11 loci. Two PCR triplex reactions were developed and optimised, combining the three new loci with established markers. Six hundred UK individuals who separately described themselves as white, black or South Asian (from the Indian subcontinent) were typed and intermediate alleles and duplications further characterised. Addition of the new loci significantly increased haplotype diversity.

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1. Introduction

Y-chromosome markers can provide additional benefits over autosomal STRs, for example, assisting in complex relationship studies and providing additional and more

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sensitive information about individuals involved in an allegation of rape. Because of generally low levels of polymorphism, leading to poor individual discrimination, and the inherent linkage between the markers, profiles must be analysed as haplotypes, rather than as independent loci. We wished to increase the number of markers we use, adding three new markers [1] to the standard eight (DYS 19, 385, 389-I, 389-II, 390, 391, 392, 393) to improve discrimination.

2. Materials and methods

2.1. Six hundred male individuals were typed from the three ethnic groups most prevalent in the UK: Caucasians and Afro-Caribbeans and South Asians

Donors comprised mainly of individuals sampled for paternity analysis from mainland Britain supplemented by historic and ongoing collections of unrelated individuals. All donors provided consent and volunteer donors were made anonymous on collection for further protection. DNA was obtained from blood samples or mouth swabs and extracted using a standard Chelex method.

2.2. Three multiplexes (pentaplex, triplex 1 and triplex 2) were used to generate dye labelled products from the eleven loci

An existing and widely used pentaplex combination amplified the loci: *DYS* 19, 389-I/II, 390, and 393 [2]. Triplex 1 comprised *DYS*391, 437, and 439, which were amplified under the following conditions: 95 °C 15 min, then 94 °C 1 min, 60 °C 1 min, 72 °C 1 min using TouchDown PCR with eight cycles, each reducing the annealing temperature by 0.5 °C, followed by 22 cycles of 94 °C 1 min, 56 °C 1 min, 72 °C 1 min, ending with 72 °C 5 min. Primer concentrations were: *DYS*391 0.25 µM, *DYS*437 0.4µM, and *DYS*439 0.25µM using 2 ng of DNA. Triplex 2 comprised *DYS*385, 392, and 438, which were amplified under the following conditions: 95 °C 15 mins then 94 °C 1 min, 60 °C 1 min, 72 °C 1 min using TouchDown PCR with eight cycles, each reducing the annealing temperature by 0.5 °C, followed by 30 cycles of 94 °C 1 min, 56 °C 1 min, 72 °C 1 min, ending with 72 °C 5 mins. Primer concentrations were: *DYS*385 0.2 µM, *DYS*392 0.5 µM, and *DYS*438 0.3 µM using 2 ng of DNA. Allelic ladders were constructed for the three new loci, and all components were sequenced to confirm repeat number and absence of sequence anomalies.

3. Results

3.1. Locus diversity varied from a low of 0.28 (*DYS*392 in Afro-Caribbeans) to a high of 0.95 (*DYS*385 in Afro-Caribbeans)

Haplotype diversities for the loci were all >0.999.

3.2. Adding additional markers increased the proportion of distinct haplotypes observed by 11%, to 92% in the Caucasian population, and by 5% each to 96% and 93% in the Afro-Caribbean and South Asian populations, respectively

There were 29 shared haplotypes. Family names were available for 20 of these and shared only between one pair. Five haplotypes were shared between individuals from the separate Caucasian and Afro-Caribbean populations and one haplotype between individuals from the Caucasian and South Asian populations. There was no sharing observed between the Afro-Caribbean and South Asian groups.

3.3. Intermediate alleles were seen in 5/600 individuals: *DYS19* (15.2), *DYS389-II* (29.2), *DYS385* (11–13.2), (14.2–18) and (14–16.2)

Duplications were seen in 2/600 individuals: *DYS389-I* and *-II* (13, 14 and 29, 30) seen in one individual, and *DYS385* (11–14, 11–15). All anomalies were confirmed by repeat analysis and subsequent sequencing.

4. Discussion

Like autosomal STRs, intermediate alleles are sometimes observed and in this study were most often seen in the *DYS385* paired allele locus. Duplications are seen at a similar frequency and, because many of the loci are closely linked, if observed, may be seen at more than one locus within an individual. Haplotype diversity is very high and across race group sharing is seen more often between Caucasians and Afro-Caribbeans than between Caucasians and South Asians, reflecting the social structure within the British population.

References

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