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The beneficial effect of extending the Y chromosome STR haplotype

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Abstract. Seven recently discovered Y chromosome STR markers have been investigated; DYS485, DYS499, DYS536, DYS546, DYS557, DYS570 and DYS576. The markers were successfully combined into one multiplex PCR reaction and 92 British Caucasian individuals were studied. Loci diversity values are presented here and most show high variability compared with established Y-STR markers. Haplotype diversity increases from 0.9988 to 1 when these seven loci are used to extend the 'PowerPlexY' haplotype. Also detailed is a relationship case that demonstrates the advantages that additional informative Y chromosome loci can confer when used in forensic casework.

Keywords: Y chromosome; STR polymorphism; Haplotype

1. Introduction

Y chromosome STR testing is becoming a more frequently used forensic technique both in criminal and relationship cases. One current drawback with this method is that haplotypes (profiles) can be identical between non-related individuals—they are not unique.

During the development of Y chromosome testing there was a scarcity of known Y-STR loci and it is only recently that larger numbers of polymorphic markers have been discovered. Haplotype diversity is therefore partially compromised in standard forensic Y chromosome testing protocols by the need to use the established markers, some of which exhibit low polymorphism. Discrimination can therefore be increased by analysing either a

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larger set of Y chromosome markers, or by using different markers with greater polymorphism.

We have taken 7 of the new Y-STR loci recently described [1,2] and purported to be highly polymorphic. The aim was to characterize the variability of these markers on a set of individuals from the British population, assessing both the relative polymorphism and the effect on haplotype diversity.

2. Methods

Candidate Y-STR markers were obtained from the literature [1,2] on the basis of polymorphism. The sequences associated with these STR loci were downloaded from NCBI via a BLAST search (http://www.ncbi.nlm.nih.gov/BLAST/) and primers designed with the intention of producing one multiplex to amplify all loci. Markers were discarded if the sequences prevented effective multiplex primer design. Forward and reverse primers were successfully designed for 7 markers: DYS485, DYS499, DYS536, DYS546, DYS557, DYS570 and DYS576; and the PCR was optimized so that all 7 loci would co-amplify in one reaction.

This 7 marker multiplex was used to profile 92 unrelated British Caucasian males sampled in the first instance for paternity testing. These samples had previously been analysed with the markers DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438 and DYS439; those loci present in the PowerPlex Y kit (Promega).

Reactions were carried out in 10 μl volumes, with the addition of 1ng of Chelex™ extracted DNA and 0.5 U AmpliTaq Gold DNA polymerase (Applied Biosystems). Products were separated, detected and sized by capillary electrophoresis on an ABI PRISM™ 3100 Genetic Analyser in combination with Genescan™ 3.1 analysis software.

Table 1
Gene diversity values for all 18 Y-STR markers in a British caucasian population

Locus	Gene diversity
DYS385	0.822
DYS449	*0.790*
DYS570	*0.761*
DYS576	*0.753*
DYS546	*0.732*
DYS389II	0.704
DYS390	0.700
DYS439	0.651
DYS557	*0.593*
DYS392	0.574
DYS438	0.553
DYS437	0.542
DYS391	0.519
DYS389I	0.518
DYS485	*0.503*
DYS19	0.481
DYS536	*0.460*
DYS393	0.350

Allelic ladders were constructed and sequenced to allow allelic determination. Locus and haplotype diversity values were calculated [3] for the population.

3. Results

All of the markers proved to be polymorphic in the population studied, in particular DYS449, DYS570, DYS576 and DYS546 showed higher levels of variance compared with the established markers (with the exception of the biallelic marker DYS385). Gene diversity values are detailed in Table 1 for all 18 markers.

The addition of these extra 7 Y-STR loci to the 11 markers already present in the PowerPlexY kit, can be seen to increase haplotype discrimination. Of the 92 samples profiled, with the 11-marker analysis 88 different haplotypes were observed giving a haplotype diversity of 0.9988. When all 18 markers were analysed it was possible to differentiate all 92 samples, and the previously shared haplotypes were separated at a minimum of 4 of the new loci, demonstrating the utility of these extra markers.

4. Discussion

A number of the loci originally identified as targets from the literature had to be discarded due to the condition of the sequence surrounding the STR which made efficient multiplexing difficult. This may mean that some of the more promising of the recently discovered markers may not prove to be as valuable as anticipated.

The seven new STRs that were examined proved to be very useful in discriminating between unrelated individuals who previously shared the same 11 marker haplotype. All shared haplotypes were variations on the ‘most common European haplotype’, so the variation in these new single allele markers is very encouraging. Some of these markers prove to be so polymorphic in comparison to the established Y-STR loci that consideration should be given to incorporating these markers into regular Y chromosome analysis.

Addition of the extra markers not only helps individualisation of samples, but has also proved useful in some complex paternity cases where there are 1 or 2 inconsistencies (exclusion or mutation). In one such case, 2 men wanted to discover if they were full or half siblings. Autosomal testing showed that they were 400 times more likely to be full brothers but there was one Y chromosome exclusion. Addition of the extra Y-STRs produced no additional mis-matches supporting the view that the original Y difference was most likely a mutation.

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