Short communication

The impact of additional Y-STR loci on resolving common haplotypes and closely related individuals

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Abstract

Commercial Y-STR kits have permitted laboratories to go beyond the original nine minimal haplotype loci (MHL) and to discover the advantage of additional Y-STR loci in resolving common haplotypes. In an effort to examine the impact of Y-STR markers beyond the 17 loci now available in commercial kit form, new Y-STR loci are being investigated on a common set of samples representative of the major U.S. population groups. Additional Y-STRs can also increase the power of discrimination between closely related male individuals, which is important not only in forensics but also in the paternity and genetic genealogy communities.

Keywords: Short tandem repeat; STR; Y-chromosome; Y-STR; DYS449; DYS570; DYS576; DYS534; NIST

1. Introduction

Y-chromosome STR analysis is a valuable tool to add to the arsenal of techniques available for human identification. In forensics, Y-STRs prove useful in sexual assault situations where there is little male DNA in the presence of high amounts of female DNA. Paternity and genetic genealogy laboratories are discovering the value of adding new Y-STR loci beyond what is commercially available in an attempt to resolve closely related individuals such as fathers and sons and for tracing paternal lineages. Therefore it is necessary to evaluate these loci to determine their value and possible future adoption by commercial kit providers and the genetic genealogy community.

Two commercial Y-chromosome STR kits are widely available for amplifying 12 [1] or 17 [2] Y-STR loci. Using these commercial kits as a starting point, additional loci were screened to determine their ability to resolve U.S. population samples. A primary evaluation of 27 Y-STR loci (including DYS635 in the Yfiler kit) and their allele frequencies was reported previously [3]. After testing, 20 of these loci were evaluated in conjunction with the Yfiler loci on their ability to resolve the most common haplotype observed in 656 U.S. population samples. With the addition of these loci, all samples with the most common type were able to be completely resolved.

2. Materials and methods

A total of 656 male U.S. population samples were typed with the 37 loci listed in Table 1. The Yfiler kit contains the loci in bold. The GenBank accession for each locus is listed along with the PCR product size and the location of the locus on the Y-chromosome.

The loci evaluated were either from the Yfiler kit [2] or were selected based on best candidates from previous studies [4,5]. Using the available Genome Database (GDB) primers (http://www.gdb.org), a BLAST search was conducted to locate the sequence region containing the loci. The Y-chromosome locations were determined using BLAT (http://genome.ucsc.edu/cgi-bin/hgBlat) and the updated March 2006 human
genome reference sequence. The loci were then divided into small multiplexes of four or five loci based on size and tested with population samples to determine allele ranges and gene diversity values. Conditions for PCR amplification, electrophoresis, typing and sequencing were as previously described [3].

3. Summary

We have typed over 656 U.S. population samples with 37 Y-STR loci, 17 of which are commercially available in the Yfiler kit along with 20 additional Y-STRs. Typing of these samples revealed a common haplotype that was observed 26 times with only the minimal haplotype loci [6]. Therefore about 4% of the samples were unresolved using these nine loci. Adding DYS438 and DYS439 loci [6] breaks these samples into three groups. The additional locus DYS437 in the Powerplex Y kit further resolves the samples into seven groups. The Yfiler kit adds five more loci, separating all but three samples with the most common type. Adding either DYS522 or DYS576 to the Yfiler loci resolves all 26 samples with the most common type. In this population set, 7 of the 20 additional Y-STRs have the same ability to resolve sample haplotypes as all 20 loci and therefore demonstrate promise for future multiplexing. These loci include DYS449, DYS505, DYS522, DYS532, DYS534, DYS570, and DYS576. Future studies will evaluate other Y-STRs beyond these 37 loci that may also be useful to Y-STR testing communities.

References


