



B37 Examination of Additional Y-STR Loci for Increased Resolution of Common Haplotypes

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After attending this presentation, attendees will have a method for examining new Y-STR loci in order to determine an optimal set of markers to use for increased resolution of common haplotypes and also for distinguishing between related males.

This presentation will impact the forensic community and/or humanity by demonstrating methodologies used for examining new Y-STR loci.

The need for more Y-STR loci has become increasingly important as the potential forensic uses of Y chromosome testing are revealed. One major challenge in forensic investigations has been the recovery of genetic information from perpetrators in sexual assault cases involving low amounts of male DNA mixed with high levels of female DNA. While several commercial Y-STR kits have been developed to focus on this issue, additional loci could assist in increasing the power of discrimination between closely related male lineages. At the same time, new Y-STR loci may also help to separate related males, such as fathers and sons.

We have examined 27 Y-STR loci across approximately 660 U.S. Caucasian, African American, and Hispanic samples (1). A guide for evaluating these loci will be described including examples of the distribution of allele frequencies among these populations and their gene diversity values. In addition, a subset of these Y-STR loci is in full concordance with previous studies using in-house multiplex assays and commercial Y-STR kits.

Potential new Y-STR loci were selected from the 166 new Y-STR loci described by Kayser et al. (2). The loci were mapped using PCR primers present in the Genome Database (GDB) (3). Primer pairs were checked in sets of three for primer-dimer and hairpin structures using the AutoDimer program (4). The loci were combined into 5 multiplex sets and characterized for all 660 population samples. From this information, at least two alleles for each Y-STR locus were chosen for sequencing in order to determine repeat number and to assist in nomenclature decisions. Once the repeat number was established, allele frequencies were determined for the loci. Gene diversity values for the individual loci in the population samples were examined as well as a collection of haplotype information.

Twenty-seven (27) additional Y-STR loci have been studied and characterized and at least 20 of these have been found to have high variation and genetic diversity values in three major U.S. population groups.

An approach for looking at new Y-STR loci has been established. Future studies will investigate other Y-STR loci that may be of forensic value to potentially supplement commercially available Y-STR loci kits. These loci will also be evaluated on their ability to distinguish between father/son pairs.

References:

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2. Kayser, M.; Kittler, R.; Ralf, A.; Hedman, M.; Lee, A.C.; Mohyuddin, A.; Mehdi, S.Q.; Rosser, Z.; Stoneking, M.; Jobling, M.A.; Sajantila, A.; Tyler-Smith, C.; A Comprehensive Survey of Human Y-chromosomal Microsatellites, *Am. J. Hum. Genet.* 74 (2004) 1183-1197.
3. GDB; <http://www.gdb.org>
4. Vallone, P.M.; Butler, J.M. AutoDimer: A Screening Tool for Primer-dimer and Hairpin Structures, *Biotechniques* 37 (2004) 226-231.

Short Tandem Repeat, Y-Chromosome, DNA