B156 Y-Filer and Beyond: Mutation Rates and New Loci for Increased Haplotype Resolution

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The audience will become familiar with a set of Y-STR loci that can be used beyond commercially available loci for further haplotype resolution and will also be provided with updated information on mutation rates with the 17 Y-STR loci in the Yfiler kit.

This presentation will impact the forensic community by introducing new multiplex of Y-STR loci that will allow for further resolution of sample haplotypes beyond commerically available kits.

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An important part of studies with the Y chromosome is to determine Y-STR loci that are highly discriminatory in order to extract maximum informative value from a sample. The focus of this work was to determine an optimal set of Y-STR loci that can be used for further resolution of male individuals beyond those loci contained in commercially available kits. Close to 100 Y-STR loci were examined with 95 U.S. population samples and those that provided the highest discrimination were combined into a multiplex. These Y-STR loci are being tested with high levels of female DNA to ensure that they have no cross-reactivity with the X chromosome, which would make this multiplex attractive for sexual assault cases.

Another added benefit of these additional Y-STRs is their ability to increase the power of discrimination between closely related male individuals, such as fathers and sons. Currently, we have obtained 400 father:son sample pairs from Caucasians, African Americans, Hispanics, and Asians. Mutation rates using the 17 Y-STR loci in the Yfiler kit with these samples will be discussed as well as a comprehensive summary of mutation rates from literature. Mutations observed with Y-STR loci beyond those in Yfiler will also be investigated.

Methods and Materials: Potential new Y-STR loci were selected from the 166 Y-STR loci described by Kayser et al[1] as well as other various publications. The loci were mapped using PCR primers present in the Genome Database (GDB).[2] Primer pairs were checked for primer-dimer and hairpin structures using the AutoDimer program.[3] The loci were evaluated in singleplex and then combined into multiplexes of 3 or 4 loci and characterized for 95 U.S. population samples. These loci were also tested with 300ng of female DNA to ensure there was no binding on the X chromosome. Gene diversity values for the individual loci in the population samples were examined as well as a collection of haplotype information.

Summary of Results: Additional Y-STR loci have been studied and characterized and a subset of these have been combined into a multiplex that can provide Y chromosome specific results in the presence of a high amount of female DNA.

Conclusions: A multiplex of Y-STR loci that can provide increased discrimination capacity beyond the Y-STR's currently available in commercial kits has been created. Future studies will involve testing these Y-STR loci with father/son pairs.

References:

1 M. Kayser, R. Kittler, A. Ralf, M. Hedman, A.C. Lee, A. Mohyuddin, S.Q. Mehdi, Z. Rosser, M. Stoneking, M.A. Jobling, A. Sajantila, C. Tyler-Smith, A comprehensive survey of human Y-chromosomal microsatellites, Am. J. Hum. Genet. 74 (2004) 1183-1197.

2 GDB; http://www.gdb.org.

3 P.M. Vallone, J.M. Butler, AutoDimer: a screening tool for primer- dimer and hairpin structures, Biotechniques 37 (2004) 226-231.

Y-Chromosome, Short Tandem Repeat, Mutation Rates

 ** Presenting Author*