A128 Examination of Rapidly Mutating Y-STR Loci for Increased Resolution of Common Haplotypes

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After attending this presentation, attendees will be introduced to a set of rapidly mutating Y-STR markers. Population statistics on the specific loci examined and their utility for discriminating closely related males will be presented.

This presentation will impact the forensic science community by introducing the usefulness of a set of rapidly mutating Y-STR loci to increase discrimination among closely related males.

Y-chromosomal STR (Y-STR) testing has become an important tool for forensic investigations, especially in DNA mixture samples where low level male DNA is mixed in a high female DNA background. Presently, two commercial kits of 17 and 23 Y-STR markers are available for the forensic community and provide relatively high discrimination among unrelated individuals with a discrimination capacity greater than 97% (NIST, unpublished data).[1,2] Given the haploid nature of the Y-chromosome, a match between the evidence and the accused is evaluated in terms of how frequently the haplotype is observed in a relevant database. In addition to the limitation of Y-STR statistical results restricted to the size of the database, the current set of Y-STR markers is limited at separating related males such as fathers and sons and brothers.

Recently published rapidly mutating (RM) Y-STR loci with mutation rates from roughly 1 to 7% per meioses were evaluated at NIST using a set of unrelated population samples to determine population genetics parameters such as haplotype diversity and among a set of Father-Son samples to determine the usefulness of the markers for distinguishing related males.[3,4]

Fifteen RM Y-STR markers were organized in three multiplex reactions and amplified according to Ballantyne *et al.*[3] NIST population samples of over 600 unrelated individuals in three U.S. groups: Caucasian, African American, and Hispanics were initially tested.[5] Nearly 400 father-son samples among U.S. Caucasian, African American, Asian, and Hispanics were also tested.[6] Previously all samples have been typed for the two commercially available forensic Y-STR kits.

The study found that the RM Y-STR markers provided increased discrimination and variation among common haplotypes unresolved from the two commercially available Y-STR kits. For the father-son samples, over 21% of the samples tested exhibited at least one mutational event among the RM Y-STRs.

Conclusions: Additional Y-STR loci, especially from rapidly mutating markers, can be useful for increased discrimination among closely related males.

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