



B182 Examination of Rapidly Mutating Y-STR Loci for Increased Resolution of Common Haplotypes Using a Large Multiplex Kit

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The goal of this presentation is to introduce attendees to a set of rapidly mutating Y-chromosomal Short Tandem Repeat (Y-STR) markers in a commercially available multiplex kit. 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 in a commercially available kit to increase discrimination among closely related males.

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. Until 2014, only two commercial kits of 17 and 23 Y-STR markers were available for the forensic community that provide relatively high discrimination among unrelated individuals, with a discrimination capacity greater than 97% (National Institute of Standards and Technology (NIST), unpublished data).^{1,2} Given the haploid nature of the Y-chromosome, a match between the evidence and the accused is evaluated in terms of how frequently the haplotype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 are limited at separating related males such as fathers, 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.^{3,4} To assess the utility of RM Y-STR markers for casework, NIST has evaluated the Yfiler® Plus kit released for commercial use in 2014. The Yfiler® Plus kit includes seven rapidly mutating markers intended to increase discrimination among closely related individuals. The evaluation used a set of unrelated population samples to determine population genetics parameters, such as haplotype diversity, and a set of father-son samples to determine the usefulness of the markers for distinguishing related males.

NIST population samples of more than 600 unrelated individuals in three United States groups, Caucasian, African American, and Hispanic, were initially tested with the Yfiler® Plus kit.⁵ Subsequently, nearly 400 father-son samples among United States Caucasians, African Americans, Asians, and Hispanics were also tested.⁶ All samples had been previously typed using the original two commercially available forensic Y-STR kits.^{1,2}

As a result of the testing, NIST found that the RM Y-STR markers provided increased discrimination and variation among common haplotypes unresolved using the original commercially available Y-STR kits. Additional Y-STR loci, especially from rapidly mutating markers, can be useful for increased discrimination among closely related males; however, caution should be considered in the interpretation of these markers when discrepancies occur among close relatives that could lead to incorrect exclusions based upon a pre-determined number of mismatches (e.g., in missing person cases).



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