



A132 Development Criteria for a Next Generation Y-STR Multiplex for Forensic Applications

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After attending this presentation, attendees will understand different aspects and challenges in designing a Y-Chromosome Short Tandem Repeat (Y-STR) multiplex that can be used for multiple forensic and human identification applications.

This presentation will impact the forensic science community by demonstrating how loci selection in a multiplex might favor one application over the other. On the other hand, if enough loci are multiplexed, it might be possible to apply subsets of these loci for specific applications.

Y-STRs have been used in human identification for over a decade in paternity testing, male lineage studies, and forensic DNA analysis. Y-STRs offer certain advantages over autosomal STR analysis, such as determining the number of male contributors in a sample, multigenerational male lineage studies, and the ability to obtain a male profile in the presence of a high background of female DNA bypassing the need for performing differential extraction of sperm and epithelial cells. Y-STRs can be used for rapid exclusion of suspects. However, the discrimination power of Y-STRs is lower than autosomal STRs and non-exclusion cases may need further investigation.

In recent years, population data for several new Y-STR loci have become available in the published literature. Some of these Y-STR loci are highly discriminating and thus offer good potential for use in forensic DNA analysis. Selection of the Y-STR loci for constructing a multiplex is critical and determines its downstream applications. If the objective is to exclude close patrilineal relatives of the suspect, then markers with a high mutation rate are preferred. On the other hand, in kinship analysis, markers with high mutation rates might prove problematic. In designing a new forensic Y-STR multiplex, the inclusion of currently used Y-STRs should be considered, given that the existing Y-STR databases are already populated with profiles containing this information. New markers could be added to enhance the capabilities of already existing Y-STR multiplexes. These enhancements could combine features such as mini-STRs, the inclusion of highly discriminating markers which could allow for better differentiation of paternal lineages in populations with low Y-chromosome diversity and rapidly mutating markers.^{1,2} In addition, a next generation kit should also provide improved performance for profiling of challenging samples when compared to already existing multiplexes. These enhancements include improvements to the overall amplification balance, improved resistance to inhibitors of the PCR, and shorter amplification times.

This presentation will discuss a strategy for the development of an enhanced Y-STR multiplex that combines well-known loci as well as recently characterized, highly discriminating Y-STRs into a single amplification reaction. The presentation will also address how different nomenclatures could lead to potential lack of concordance with already existing published reports in some Y-STRs. Results will show that full profiles are attainable with low levels of male DNA (below 150 pg) and that under optimized conditions no detectable cross-reactive products were obtained from human female DNA, bacteria, and commonly encountered animal species. Additionally, results demonstrate the ability to detect male-specific profiles in admixed male and female samples at ratios greater than 1:1000. The haplotype diversity and discriminatory capacity calculations for the three major U.S. population groups (Caucasians, Hispanics, and African Americans) will also be described.

References:

1. Ballantyne KN, Keerl V, Wollstein A, Choi Y, Zuniga SB, Ralf A, Vermeulen M, deKnijff P, Kayser M. *A new future of forensic Y-chromosome analysis: Rapidly mutating Y-STRs for differentiating male relatives and paternal lineages*. Forensic Sci Int Genet 2012;6(2):208-18.
2. Ballantyne KN, Kayser M. *Additional Y-STRs in forensics: Why, which, and when*. Forensic Sci Rev 2012; 24(1): 63-78.

Y-STR, Forensic Casework, Paternity