B42 Microhaplotypes for Kinship Analyses

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Learning Overview: After attending this presentation, attendees will understand the potential application of Microhaplotype (MH) biomarkers in kinship analysis.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by proposing MHs as a supplemental tool to conventional Short Tandem Repeat (STR) typing analysis for kinship inference.

MHs are emerging biomarkers of at least two Single Nucleotide Polymorphisms (SNPs) associated in multiple allelic combinations within 300bp. The multi-allelic nature of MHs make them more informative than a Single Nucleotide Polymorphism (SNP) locus and useful for forensically relevant applications, including mixture deconvolution and ancestry inference on massively parallel sequencing platforms. In addition, they may prove helpful in kinship inference due to their low mutation rate. However, no study has fully demonstrated the potential of these markers in kinship inference. This study aimed to fill this gap by testing the utility of MHs in paternity and kinship testing by testing different scenarios.

An initial set of 347 individuals from four United States population groups (88 Afro-American [AA], 114 European-American [EA], 102 Southwest Hispanic [His], and 43 East-Asian American [EAA]) was genotyped using a 74 MH bioassay on the Ion S5™ System sequencing platform.

Allele frequencies for each population were calculated. A total of 1,000 simulation tests were performed for each population to determine Likelihood Ratio (LR) thresholds under the following four kinship scenarios: parent-child, full-siblings, half-siblings, and cousins. To achieve this, the commonly used and forensically relevant Familias software v. 3.2.8 was tested to take MH data.

Overall, the LR distribution median values ranged from 10^{14} to 10^{12} for parent-child pairs, from 10^{10} to 10^{11} for full-siblings pairs, and from 10^{2} to 10^{3} for half-sibling pairs. The distribution of LR median values for cousin's was found to be approximately equal to one, thus suggesting the need to use a larger marker assay to better infer this kinship scenario.

Subsequently, a total of 1,000 simulation tests were performed on 29 autosomal STRs using allele frequencies from the National Institute of Standards and Technology (NIST) dataset. The distribution of LR median values for parent-child and full-siblings was found to be approximately equal to 10^{9} and 10^{7}, respectively, while for half-siblings and cousins, both were approximately equal to one.

Overall, the high LR values obtained using the 74-MH bioassay, in comparison with LR values achieved with commonly used autosomal STR markers, supports the effectiveness of these new loci in kinship inference. In addition, the ability to distinguish close (parent-child and full-siblings) and more distant (half-siblings) familial scenarios highlights the possibility of solving complex family pedigrees, which deserves further investigation.

Microhaplotypes, Kinship Inference, Likelihood Ratios

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