

MINING THE MINOR CONTRIBUTOR OF SIMULATED TOUCH DNA MIXTURES: DIP-STR VS AUTOSOMAL STR AND Y-STR SETS

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Within a forensic context, autosomal STRs (a-STRs) represent the mainstay of forensic DNA markers due to their high discriminating power. However, their capability to detect the minor DNA contributor of two- source DNA mixtures is limited to moderately unbalanced mixtures. Y-STRs can be used when dealing with unbalanced mixtures. The drawbacks of these markers are the inability to discriminate between paternally-related individuals and the required specific gender mismatch. Indeed, Y-STRs can only be used when the major contributor is a female and the minor a male. To overcome those limitations an innovative, highly sensitive and not gender-related compound marker, known as DIP-STR, was recently developed [1]. In this study we aimed at comparing the performance of a-STR, Y-STR and DIP-STR markers to target the minor contributor of forensically relevant two-source contact stains.

To address this objective we designed a specific mock case scenario-based protocol, involving 14 volunteers. Participants were randomly chosen to act either as object's owners or second users, respecting an even gender combination pair. Object's owners were asked to use regularly selected plastic- and metal-made items throughout a 10-day period; the second users then grabbed the object for 5 to 120 minutes. A first series of 132 mixed-contact stains were simulated and DNA profiles established using the NGM SElectTM, PowerPlex® Y23 commercially available kits and a set of 6 DIP-STRs.

According to the a-STRs, 6, 40 and 86 single-, two- and multi-source DNA profiles were obtained, respectively. Apart from 17 cases, the object owner was always the major contributor of the a-STR DNA profiles. A-STRs succeeded to detect the complete minor DNA component's profile in 49% of two-person mixtures. However, they were conversely inconclusive in single- and complex mixed-profiles. Y-STRs, applicable to 46% of samples, those with the required gender mismatch, detected full minor DNA's profile in 31% and 46% of two- and multi-source mixtures, but failed on the few single-person profiles. The amplification of variable subsets from 1 to 4 informative DIP-STRs showed a successful rate of 80%, 75% and 83% for simple-, two- and multi-source a-STR DNA profiles, respectively. Despite the limited number of DIP-STRs amplified, the performance of this marker was globally enhanced in low template and extremely unbalanced mixtures compared to a-STRs and Y-STRs. This was due to increased stutter peaks, allele drop-out and drop-in events, masking effect of the major DNA. More data will be presented in the poster.

Our preliminary results suggest that DIP-STRs represent a reliable supplementary and confirmatory typing tool for solving challenging forensic caseworks. Further simulations which include additional substrate-types are currently ongoing to widen the number of mock contact stains to be investigated.

Reference

[1] V. Castella, J. Gervaix, D. Hall, DIP-STR: highly sensitive markers for the analysis of unbalanced genomic mixtures, *Hum. Mutat.* 34 (2013) 644–654.