B129 The Evaluation of the Short Tandem Repeat (STR) Genotype Concordance Between Massively Parallel Sequencing (MPS) and Capillary Electrophoresis (CE) STR Kits in Japanese Population Samples

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Learning Overview: After attending this presentation, attendees will understand the performance of Short Tandem Repeat (STR) typing kits by Massively Parallel Sequencing (MPS) and Capillary Electrophoresis (CE). Attendees will also understand the concordance rates comparing allele calls from MPS and CE STR kits in Japanese population samples.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by reporting the concordance rates and sources of discordant of STR allele calls between MPS and CE STR typing kits.

In this study, the STR profiles were generated and compared for the purposes of generating population allele frequencies and understanding allele call concordance by both MPS and CE methods. For the population study, buccal swab samples were collected from 83 unrelated Japanese volunteers, and DNA was extracted using EZ1 Advanced XL with EZ1 DNA Investigator Kit. For the allele call concordance study, the 83 samples mentioned above and 39 samples selected from a larger set of 1,501 samples previously extracted from unrelated Japanese volunteers were used. Out of the selected 39 samples, 30 contained D19S433 silent allele (silent: GlobalFiler® PCR Amplification Kit; normal: PowerPlex® Fusion System).[1]

The MPS STR profiles were generated by Precision ID GlobalFiler® NGS STR Panel v2 with the Ion S5/S5XL/Chef System and ForenSeq DNA Signature Prep Kit with MiSeq FGx Forensic Genomics System and compared with the CE STR profiles generated by the GlobalFiler® PCR Amplification Kit and PowerPlex® Fusion System analyzed on a 3500xL Genetic Analyzer. The CE STR data from GlobalFiler® PCR Amplification Kit and PowerPlex® Fusion System were analyzed using GeneMapper® ID-X 1.4 software. The data from Precision ID GlobalFiler® NGS STR Panel v2 were analyzed using Converge Software v2.0 and HIDGenotyper plugin v2.0. The data from the ForenSeq DNA Signature Prep Kit were analyzed using ForenSeq Universal Analysis Software and STRait Razor v2s.

For population samples, there was no discordance observed in the allele calls generated by ForenSeq DNA Signature Prep Kit and CE STR kits except higher stutter and allele dropout caused by low peak height ratio found at some loci when using the ForenSeq DNA Signature Prep Kit. There were 6 discordances in 6 individuals at D2S441 locus between the Precision ID GlobalFiler® NGS STR Panel v2 and CE STR kits caused by a 1 base pair insertion just upstream of repeat region (9: Precision ID GlobalFiler® NGS STR Panel v2; 9.1: CE STR kits). There were 41 apparent discordances in 36 individuals at D10S1248, D12S391, and D1S1677 loci between the Precision ID GlobalFiler® NGS STR Panel v2 and CE STR kits that arose from bioinformatics issues found in the software and plugin.

The silent alleles at D19S433 were not amplified with Precision ID GlobalFiler® NGS STR Panel v2 as with the case of GlobalFiler® PCR Amplification Kit. On the other hand, the same "silent" allele at D19S433 was successfully amplified with ForenSeq DNA Signature Prep Kit as well as the PowerPlex® Fusion System.

Overall, MPS STR typing kits produced more discriminative information compared with CE STR typing kits. However, there were some bioinformatics issues on the reliability of the MPS STR analysis software that need to be resolved to produce full concordance to the known CE STR allele calls. The software updates of the algorithm may correct the discordances reported here in the near future.

Reference(s):

1. Fujii, K. et al., Typing concordance between PowerPlex® Fusion and GlobalFiler® based on 1501 Japanese individuals and the causes of typing discrepancies., *Forensic Sci Int Genet*., 2016; 25:e12–e13.

Massively Parallel Sequencing, Short Tandem Repeat, Concordance