



Criminalistics Section - 2016

B2 The Effectiveness of Various Strategies to Improve DNA Analysis of Formaldehyde-Damaged Tissues From Embalmed Cadavers for Human Identification (HID) Purposes

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After attending this presentation, attendees will better understand the effectiveness of various strategies to improve DNA typing from severely damaged and degraded Formalin-Fixed (FF) tissue samples for HID purposes.

This presentation will impact the forensic science community by suggesting that, rather than attempting to improve the quantity and quality of severely damaged and degraded DNA template (present in FF samples) prior to Short Tandem Repeat (STR) typing, a more productive approach for HID purposes may be to utilize Insertion/Deletion (INDEL) panels or Single Nucleotide Polymorphism (SNP) markers using Massively Parallel Sequencing (MPS) technologies.

FF tissues for genetic analysis may be obtained from autopsied or archived pathology samples, embalmed cadavers, or repatriated remains. STRs have been the gold standard markers for DNA HID for more than 15 years; however, after treatment with formalin fixatives, many samples are not successfully genotyped using STR analysis. Instead, other methods that pre-amplify the low amount of good quality DNA, repair the damaged DNA template, or use alternate genetic markers to amplify smaller target regions may generate more probative genetic information from these samples.

This study investigated the ability of three different Whole Genome Amplification (WGA) methods (GenomePlex® Complete WGA Kit; Illustra™ Ready-To-Go™ GenomiPhi™ V3 DNA Amplification Kit; and QIAGEN® REPLI-g FFPE Kit) plus one DNA repair treatment to improve downstream STR typing of FF tissues from embalmed cadavers.

In addition, the use of bi-allelic markers, such as INDELs and SNPs, were investigated. These markers, smaller than 200 base pairs (bp) in length, are less susceptible to degradation and therefore may also be more likely to amplify in highly damaged DNA, as in the case of FF tissues. The comparative Random Match Probabilities (RMP) of each sample using STRs, a battery of 39 INDEL markers, and a panel of 124 SNP markers using MPS for HID purposes (HID-Ion AmpliSeq™ Identity Panel) were examined.

This study presents the results of this work, in which none of the three WGA methods or the DNA repair treatment tested in this study consistently yielded more complete STR profiles than the untreated FF samples; however, when the RMPs of each sample obtained using the INDEL and SNP-based MPS panels were compared to those generated from the partial STR profiles obtained from non-treated FF samples, the INDEL and SNP markers generated notably lower RMPs, providing more robust DNA identifications.

Formalin-Fixed, INDELs, Massive Parallel Sequencing