APPLICATION OF ENHANCEMENT STRATEGIES FOR THE IMPROVEMENT OF FORENSIC DNA PROFILING FROM HUMAN BONES

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Even though forensic DNA profiling techniques are considered a powerful method for human identification, problems arise when low quantity and/or low quality DNA is tested. Analyzing forensic samples with low DNA template using standard genotyping techniques like short tandem repeat (STR) typing can yield no profile or an incomplete profile, which makes conclusive identification of the sample nearly impossible. An example of a challenging forensic sample is bone tissue. DNA within bone may contain potential PCR inhibitors and can also be degraded as a result of persistent exposure to harsh environmental conditions. Consequently, the amount of usable DNA in bones can be extremely limited which complicates the downstream STR applications for DNA profiling.

To achieve a more discriminating DNA profile from challenging samples such as bones, enhancement strategies may be used to improve traditional STR typing methods. Whole genome amplification (WGA) can be used to amplify low quantities of starting DNA template and results in large quantities of amplified product. Because of the increased DNA yields, WGA techniques may provide better detection and coverage for STR analysis. In addition, single nucleotide polymorphism (SNP) analysis using massively parallel sequencing (MPS) technologies may be an alternative approach to characterizing insufficient and/or degraded DNA. MPS technologies can target multiple SNPs in a single reaction, reducing the time and cost of analysis, while still maintaining a high power of discrimination. By analyzing the smaller SNP markers, it is possible to successfully obtain information from degraded DNA that is normally too fragmented to characterize by traditional STR techniques.

This study investigated techniques and emerging technologies that would improve the ability to obtain a DNA profile from human bones, which are often a challenging forensic sample. Purified DNA from a human femur, rib, and phalange were examined in this study. Analysis of DNA from each bone was performed using three different genotyping/sequencing techniques: STR amplification using the GlobalFiler® PCR Amplification Kit, whole genome amplification using the REPLI-g® Mini Kit, and massively parallel sequencing using the Ion PGM™ Sequencer with the HID-Ion AmpliSeq™ Identity and Ancestry Panels. The aim of this study was to determine which enhancement strategy will produce the most discriminating DNA profile to assist with identification from these challenging forensic samples.