B142 Examination of 20 Retrotransposable Polymorphic Insertion/Null (INNUL) Markers for Their Utility in Kinship Testing Using the Commercial Software Program LSAM

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After attending this presentation, attendees will understand the effectiveness and abilities of using the software program Laboratory Information System Applications (LISA) Statistical Analysis Module (LSAM) to successfully track and manage data and documents, calculate statistics of paternity/kinship analysis, and support direct match comparisons of profiles for the INNUL markers compared to hand-calculated statistics for paternity/kinship analysis in complete and degraded sample profiles.

This presentation will impact the forensic science community by illustrating the benefits and potential of using the LSAM software for the analysis of INNUL markers in kinship and paternity testing. Also, this study examines the impact that INNUL markers have compared to other types of markers such as mitochondrial DNA (mtDNA) or Single Nucleotide Polymorphism (SNP) when testing samples with degraded DNA.

Forensic DNA testing has proven to be a powerful tool for criminal investigations and for identifying human remains via kinship analysis for cases involving mass disasters or missing persons. Short Tandem Repeat (STR) markers most often offer the highest degree of discrimination and speed of analysis by using the Polymerase Chain Reaction (PCR) and capillary electrophoresis to generate DNA profiles. DNA testing for human remains identification is often challenging due to the presence of degraded DNA or inhibitors that affect the PCR reaction. Mini Short Tandem Repeats (miniSTRs) have been engineered to decrease the size of PCR amplicons to improve recovery of DNA fragments in the high molecular weight range of standard STR kits.[1] Additional marker systems such as SNPs, Insertion/Deletion markers (InDels), and mitochondrial DNA (mtDNA) have been successfully used for highly degraded samples.

Retrotransposable Polymorphisms (RP) are found in the human genome as RNA elements that have been reverse transcribed into specific loci as complementary DNA (cDNA) by use of retroposition.[2] RP include the Alu elements Short Interspersed Nuclear Elements (SINEs) and Long Interspersed Nuclear Elements (LINEs).[3] These elements can be in two allelic states: either they are present in an individual's DNA as an insertion or absent as a null (INNULs). One advantage for targeting these markers is the small amplicon size that can be created for each marker (approximately 60bp-125bp in size).[4] A commercially available kit, InnoTyper™ 21, containing 20 INNUL markers plus the sex determining marker, Amelogenin, is available and was used for this study.4

The allele frequencies and population genetic parameters of the markers in a set of more than 600 population samples at National Institute of Standards and Technology (NIST) were characterized first.[5] Samples with close relatives (father/mother/child trios) were then genotyped for testing. Next, individuals in the trios were artificially degraded using sonication to generate fragmented DNA. Samples were typed with the INNUL markers and a commercially available multiplex STR kit for determining the Random Match Probability (RMP).

The utility of the INNUL markers for paternity and kinship analysis were considered. The software package LSAM-LISA Statistical Analysis Module from Future Technologies Inc. was used for direct comparison statistics and to conduct pedigree statistics for the INNULs. The program offered the ability to construct pedigree scenarios for kinship analysis. The software statistics were compared to those determined by hand calculation.

It was found that the INNUL markers were able to provide additional genetic information for samples that were highly degraded. The LSAM software program provided strong support for the concordance to produce accurate calculations for kinship analysis compared to hand calculation.

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