

B111 A Concordance Study Comparing Different Amplification Chemistries and Electrophoretic Platforms for a Databasing Program

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The goal of this presentation is to present the results of an extensive concordance study and collaboration performed by the Alabama Department of Forensic Sciences (ADFS) and Marshall University Forensic Science Center (MUFSC) in order to address the State of Alabama's backlog of convicted offender samples.

This presentation will impact the forensic community and/or humanity by demonstrating that either multiplex reagent kit or instrument platform can be effectively used to process convicted offender samples. The study demonstrates that the procedures employed are robust and valid. Furthermore, this study will present recommendations for the forensic community regarding the interpretation of DNA profiles developed from convicted offender samples and their subsequent entry into CODIS in order to maximize the current searching algorithm to aid in the identification of the perpetrators of violent crimes.

Concordant results using different fluorescent-based STR (short tandem repeats) genotyping systems for the analysis and databasing of convicted offender samples are critical for accurate matching to nonsuspect casework and to link serial crimes. In order for a state to upload its database of convicted offender DNA profiles into national level of the COmbined DNA Indexing System (CODIS), each profile must be comprised of the standard 13 core STR loci. Prior to this requirement, the ADFS had generated a convicted offender DNA database with eight (8) STR loci of the 13 core STR loci. With the support of the National Institute of Justice, the Alabama Department of Forensic Sciences (ADFS) and Marshall University Forensic Science Center (MUFSC) initiated a collaborative study to conduct additional DNA testing on these convicted offender samples to develop results for the remaining five (5) core STR loci and to demonstrate concordance of the STR results obtained, regardless of the testing platform employed. The STR results submitted by MUFSC were generated using System 1, the ABI PRISM® 3100 Genetic Analyzer, the AmpF/STR® Identifiler® PCR Amplification Kit, ABI PRISM® GeneScan® Software version 3.7 and Genotyper® Software version 3.7.1 (Applied Biosystems, Foster City, CA). The STR results from samples previously processed by the Alabama Department of Forensic Sciences were obtained using System 2, the Hitachi FMBIO® II Fluorescent Imaging Device (Hitachi/Miraibio Genetic Systems, Alameda, CA), the PowerPlex® 1.1 System (Promega Corporation, Madison, WI), and FMBIO® Analysis and STaR Call ™ (Hitachi Genetic Systems). This study demonstrates that the combination of instruments, reagents, procedures, and analyses employed by both of these institutions provided concordant and accurate genotyping results for databasing purposes.

Beginning in April 2002, the MUFSC submitted 5,000 STR profiles to the ADFS using System 1 had previously been processed using System 2 by the Alabama Department of Forensic Sciences. Overall, there is incredible concordance for all 5,000 samples between the two (2) systems. Greater than 99.98% concordance is achieved when comparing the overlapping eight (8) loci present in both systems. Any differences noted have been identified and summarized into the following categories: lower resolution when using System 2; stochastic amplification; and allelic dropout. An example and respective percentages of each category will be presented.

Resolution with the capillary electrophoresis platform is much better than resolution using slab gel electrophoresis. Further, it is known that the amplification kits from various manufacturers incorporate dissimilar different primer sets which may result in differences in the amplification conditions and may lead to destabilization of the amplification and eventually stochastic effects and/or allele dropout. The frequency of stochastic effects and allele dropout is extremely low. The manufacturer of the kit with the loci exhibiting the highest frequency of allelic dropout observed in this study (i.e., D13S17 and D16S539) has subsequently modified PowerPlex® kit(s) to incorporate amplification of the null alleles. Rare observations of stochastic effects and/or allelic dropout were observed and documented at various loci in both testing systems. The searching algorithms available within the CODIS software which employ moderate stringency searches and allow for a selected number of mismatches can address any low frequency differences between testing platforms.

This concordance study demonstrates that either multiplex reagent kit or instrument platform can be effectively used to process convicted offender samples. The study demonstrates that the procedures employed are robust and valid. Furthermore, this study will present recommendations for the forensic community regarding the interpretation of DNA profiles developed from convicted offender samples and their subsequent entry into CODIS in order to maximize the current searching algorithm to aid in the identification of the perpetrators of violent crimes.

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