



A84 The Evaluation of Expert Systems for the Missing Persons Program Using Common STR Kits, Mini-STRs, and Y-STRs

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After attending this presentation, attendees will understand the significance of evaluating expert systems performances when using Y-STR and mini-STR data, especially as it applies to reference sample databasing for the Missing Persons Program.

This presentation will impact the forensic community by providing data from expert systems analyses of Y-STRs and mini-STRs, thereby increasing the scope of the Missing Persons database and the efficiency of the laboratories responsible for processing the reference samples.

In response to the Presidents DNA Initiative, the National Institute of Justice (NIJ) has implemented many programs to increase awareness, provide financial support, and develop new DNA technologies. Additionally, three national laboratories are federally funded through NIJ for the Missing Persons Program: (1) University of North Texas Center for Human Identification, (2) California Department of Justice Jan Bashinski Laboratory, and (3) Federal Bureau of Investigation. These CODIS laboratories use advanced DNA technologies to process unidentified human remains and the family reference samples from biological relatives. The resulting DNA profiles (13 core CODIS loci and/or mitochondrial DNA) are uploaded to the CODIS+mito Missing Persons Index. In this database, mitochondrial and nuclear DNA profiles from the unidentified remains can be searched against the reference profiles; identifications are made through kinship analysis testing. With several hundred-thousand missing persons cases reported each year and more than 14,000 human skeletal remains retained in medical examiners' and coroners' offices, the need for continued support and development of the Missing Persons Program is evident.

Commercially available expert systems decrease sample processing time by automatically interpreting STR data. Due to the excessive number of missing persons cases, there is a potential of twenty-thousand or more reference samples from relatives of the missing to be processed. Expert systems offer improvements in databasing efficiency for reference samples, and it is anticipated that the use of this technology will soon be allowed for uploading family reference samples to the Missing Persons database. It is further anticipated that the CODIS+mito database, which catalogs family references for the Missing Persons Program, will be accepting Y-STR and mini-STR profiles. These additional profiles are of interest because Y-STRs provide information on paternal lineages in males, and mini-STRs may produce additional information in degraded samples. Therefore, this presentation focuses on an evaluation of the performance of three expert systems when using Y-STR and mini-STR data.

The National Institute of Justice's Expert System Testbed (NEST) Project has evaluated several expert systems using NDIS approved STR kits; neither Y-STRs nor mini-STRs have been included in their reports. This study provides a side-by-side evaluation of two expert systems not included in the NEST Project, GeneMarker® HID (SoftGenetics, State College, PA) and FaSTR (Environmental Science and Research, Wellington, New Zealand). Additionally, GeneMapper® ID v3.2 (Applied Biosystems, Foster City, CA), a software program familiar to the forensic community, is included in the study. The concordance study using these three expert systems will be presented, which includes reference samples amplified with the AmpfSTR® Yfiler® and MiniFiler™ PCR Amplification Kits (Applied Biosystems) Samples amplified with AmpfSTR® Identifier® PCR Amplification Kit (Applied Biosystems) and PowerPlex®16 System (Promega Corporation, Madison, WI) are also included in this study. For each analysis, resulting allele calls and rule firings are evaluated side-by-side for concordance and comparison, respectively. All rule firings are independently investigated and the information is compiled from the cumulative results for each expert system, such as the number of peaks detected and the associated allele calls and rule firings. In addition to the concordance study, the tools, user-interface, and overall functionality of each program are evaluated. Since each expert system is unique, differences in rule firings, tools, and user interfaces are observed and delineated. This information will demonstrate that mini-STRs and Y-STRs can be analyzed using expert systems. Expert systems make accurate allele calls with increased efficiency; this increased efficiency will help Missing Persons Programs address our nation's "silent disaster," the identification of missing persons.

Expert Systems, Missing Persons Program, Mini - STRs