

THE NATIONAL INSTITUTE OF JUSTICE'S EXPERT SYSTEMS TESTBED PROJECT

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Abstract

The National Institute of Justice's Expert Systems Testbed Project, or NEST Project, was publicly introduced in June 2005. An Implementation Team and the Project Team Advisors convened in Washington, DC in May 2005 to determine the focus and direction of the NEST Project.

Tremendous progress has been made in the analysis of convicted offender samples as a result of the NIJ *Convicted Offender DNA Backlog Reduction Program*. Implementation of single amplification kits, utilization of automated instrumentation, and the outsourcing of convicted offender samples has shifted the backlog to the technical review of the generated data. The NEST Project is focused on the evaluation of three commercially available expert systems that assist analysts in the technical review of convicted offender data.

The primary goal of the NEST Project is to conduct a thorough evaluation of the commercially available software programs and report the results back to the forensic community. For the purpose of this project, an expert system is one that: a) meets the criteria defined in the National DNA Index System DNA Data Acceptance Standards Appendix B (1); b) is publicly available for purchase; c) is configurable off-the-shelf software; d) is completely housed in the purchasing laboratory's facilities; and e) does not require the knowledge of computer code for its use (see Table 1). Expert systems are computer programs and/or systems that perform at the level of a human expert, and perform it consistently. Expert systems provide the users with a flag, or an explanation of the reasoning, used to support a decision or conclusion when data pass or fail to meet predefined analysis criteria. The expert systems can provide quality scoring, or ranking, of analyzed data.

This project will document the purchasing process for each of the individual software packages, the vendor optimization of the analysis parameters, the customer service and training provided by the vendors, and the utility of the software. The three systems that have been purchased and are undergoing independent evaluation by the Implementation Team are: 1) GeneMapper® *ID* v. 3.2 (Applied Biosystems, Foster City, California) (2, 3); 2) TrueAllele® System 2 (Cybergenetics, Pittsburgh, Pennsylvania) (4); and 3) The Forensic Science Service® DNA Expert System Suite FSS-i cubed (Forensic Science Service, Birmingham, United Kingdom)¹ (5). The three expert systems will be evaluated using a set of rules (criteria). The objective of this presentation is to share the progress made to date, the results obtained from the evaluation process, and the future goals of the NEST Project.

Introduction

The National Institute of Justice's (NIJ) Office of Science and Technology has initiated a project to evaluate the commercially available expert systems designed for use by forensic DNA laboratories. Expert systems could easily be one of the most important advances for the forensic DNA databanking community since the development of higher throughput instrumentation and single amplification multiplex chemistry.

¹ On the day of this presentation, The Forensic Science Service (FSS) and the Promega Corporation entered into collaboration in the marketing of this software package. All work presented in this presentation is direct communication with the FSS, and not the Promega Corporation.

The NIJ's Expert Systems Testbed (NEST) Project is initially focused on evaluating the ability of commercially available expert systems as a mechanism for rapid, accurate technical review of convicted offender single source DNA samples for the eventual upload into the National DNA Index System (NDIS).

Significance

In the field of forensic DNA analysis, superior computer automation for data storage and retrieval of convicted offender and forensic DNA data has proven quite useful. Since the FBI's Combined DNA Index System's (CODIS) inception to August 2005, over 2.6 million DNA samples have been entered into CODIS including convicted offenders and forensic casework profiles that have aided in over 27,000 investigations and resulted in 25,100 hits². As advancing computer technology has proven its utility, expert systems are now on the horizon to support the analysis and evaluation of convicted offender and forensic DNA data destined for input into CODIS.

Expert systems will give forensic analysts the ability to rapidly analyze and review and input large numbers of convicted offender and forensic DNA profiles into CODIS. In support of the CODIS program, the NIJ *Convicted Offender DNA Backlog Reduction Program*, as well as the availability of higher throughput DNA analysis systems, has resulted in the processing of over 1.4 million convicted offender samples since FY 2000. The NIJ *Convicted Offender DNA Backlog Reduction Program* has awarded over \$47 million to state agencies for the analysis of the offender samples for CODIS which have produced 4,000 blind hits and investigative leads³. However, analysis is not yet complete on all samples.

The analysis of convicted offender and forensic DNA data requires considerable training and experience. An expert system, one that interprets data with limited human intervention, could prove to be a noteworthy advancement for the forensic DNA community. Expert systems are not novel to the law enforcement community as they have been developed for hostage-taking incidents, burglary, and murder (6).

Expert Systems

Expert systems are computer programs and/or systems that perform at the level of a human expert, and perform it consistently. Expert systems provide the users with a flag, or an explanation of the reasoning, used to support a decision or conclusion when data pass or fail to meet predefined analysis criteria. The expert systems can provide quality scoring, or ranking, of analyzed data. Furthermore, expert systems are designed to be easily modified with the approving authority of a technical administrator for advanced data analysis and interpretation.

With the advances in higher throughput workflow via the implementation of sample card punch instrumentation, robotic pipetting workstations, multicapillary instrumentation, and multiplex single amplification chemistry along with the outsourcing of convicted offender samples, the bottleneck today has shifted from sample processing to sample data review required for eventual upload into NDIS. Currently, for the submission of convicted offender profiles into NDIS, each sample profile must have two independent technical reviews and be approved by the state's CODIS administrator (7). The incorporation of expert systems into laboratory processes will reduce the time required for the data review process and expedite the submission of the reviewed profiles into NDIS.

The NEST Project is focused on an evaluation of three commercially available expert systems that assist analysts in the technical review of convicted offender data. The primary goal of the NEST Project is to conduct a thorough evaluation of the commercially available expert systems and report the results to the forensic community that will educate forensic analysts about expert systems and enable informed purchasing decisions.

² CODIS: Combined DNA Index System, data acquired September 27, 2005 at <http://www.fbi.gov/hq/lab/codis/>.

³ Data provided by the National Institute of Justice, June 2005.

Initially, six expert systems were identified and considered for evaluation: 1) GeneMapper™ *ID* Software v. 3.2 (Applied Biosystems, Foster City, California); 2) TrueAllele® System 2 (Cybergenetics, Pittsburgh, Pennsylvania); 3) TrueAllele® System 3 (Cybergenetics, Pittsburgh, Pennsylvania); 4) FSS-i³ (Forensic Science Service, Birmingham, United Kingdom); 5) OSIRIS (National Institute of Health, Bethesda, Maryland); and 6) SureLockSM ID (Myriad Genetics, Salt Lake City, Utah). Of these, it was determined that three systems met the criteria and scope listed above. The three systems that have been purchased and are currently undergoing independent evaluation by the Implementation Team are: 1) GeneMapper *ID* v. 3.2; 2) TrueAllele System 2; and 3) The Forensic Science Service® DNA Expert System Suite FSS-i³.

The expert systems currently under evaluation during this phase of the project must meet the criteria defined in the National DNA Index System DNA Data Acceptance Standards Appendix B (3). These criteria state that the expert system can be a software program or set of software programs and that it performs all of following functions without human intervention: 1) identifies peaks/bands; 2) assigns alleles; 3) ensures data meet laboratory-defined criteria; 4) describes rationale behind decisions; and 5) makes no incorrect calls. Furthermore, additional scope was added to the criteria for evaluating expert systems by the NEST Project Team. The additional scope requires that the software program or set of software programs be publicly available for purchase, be configurable off-the-shelf software, be completely housed in the purchasing laboratory's facilities; and does not require the knowledge of computer code for its use (see Table 1).

The evaluation will document experiences associated with establishing each of the expert systems in the laboratory; specifically, the Implementation Team will report on the purchasing process for each of the individual software packages, the vendor optimization of the analysis parameters, the customer service and training provided by the vendors, the utility of the software, the speed of processing, the speed for analysis, and the many flags, rules, criteria, and features available with each of the software systems.

The three systems that did not meet the criteria and scope listed above were TrueAllele System 3, OSIRIS, and SureLock ID. TrueAllele System 3 is not completely housed in the laboratory as a stand-alone system; it is a web-based program. OSIRIS is not publicly available for purchase, is not configurable off-the-shelf software, and programming knowledge is required for use of the software package. Additionally, at this time, SureLock ID is not commercially available for purchase.

Evaluation Program

In order to evaluate data most commonly generated by forensic laboratories for input of convicted offender samples into CODIS, the Implementation Team conducted a survey of the State CODIS Administrators. The results from this survey will influence the datasets evaluated by the Implementation Team. There are many systems (that is, the combination of instrument and chemistry) currently used in the forensic community. Parameters had to be defined to capture those systems that, overall, are responsible for generating the maximum amount of data destined for CODIS. In summary, in order to capture more than 80% of the data generated in public laboratories and contract laboratories, in addition to capturing the backlog of offender samples awaiting review, it was determined that the NEST Project should focus on data produced on the ABI PRISM 3100 Genetic Analyzer using the Identifiler™ PCR Amplification Kit (Applied Biosystems, Foster City, California); the ABI PRISM 3100 Genetic Analyzer using the Profiler Plus™ and COfiler™ PCR Amplification Kits (Applied Biosystems, Foster City, California); ABI PRISM 377 DNA Sequencer (Applied Biosystems, Foster City, California) using the Profiler Plus™ and COfiler™ PCR Amplification Kits; and ABI PRISM 3100 Genetic Analyzer using the PowerPlex® 16 System (Promega Corporation, Madison, Wisconsin).

At the time of the presentation, the Implementation Team had purchased the software or software systems, had received training on the individual systems, had the software packages installed, and had initiated data comparison studies. The three different software packages have been evaluated for: 1) the purchase and delivery; 2) installation and optimization; 3) training provided with the purchase of each of the software packages; and 4) some technical, analytical, and flag and rule firing for phenotyping.

Purchasing, Installation, Optimization, and Training of the Software Systems

When purchasing the software systems, the purchaser needs to recognize that each of these systems functions very differently with respect to ease of purchase and delivery, the fee structure, and confidentiality agreements. Optimization is a process by which the vendor assists the consumer in determining rule settings by evaluating datasets produced by the consumer's laboratory. The consumer laboratory shares data and the laboratory's data interpretation guidelines with the vendor so the initial software settings can be recommended. Identical datasets have been provided to each vendor. Additionally, each vendor offers a training program for its software system.

Technical and Analytical

The Implementation Team has initiated the evaluation of the software systems for speed of software application, speed of analysis process, definition of rules and flags, documentation/audit tracking, and size standard check. The time it takes to import the datasets and run the application differs for each of the software systems. After running the datasets through the different software systems, the time needed to complete the analysis of the data was evaluated. Understanding and determining the analysis rules and flags for each of the software systems is quite different. One analysis tool evaluated in all systems was the size standard check.

Figures 1, 2, and 3 display the same sample data in the three software packages. Only one image per software package is displayed even though multiple views are available. Each software package provides the appropriate flags for the data and the correct corresponding phenotype.

GeneMapper™ *ID* Software v. 3.2

With GeneMapper *ID*, the hardware is purchased separately. Minimum hardware requirements are provided; however, the consumer determines the purchase of the hardware. The laboratory only purchases the software from the vendor. The purchase of GeneMapper *ID* is straightforward. The consumer determines the number of licenses required by the laboratory and a fee structure is in place. A CD is provided, and the consumer can self-install the software. The purchase of GeneMapper *ID* is an outright purchase of the software; no confidentiality agreements are required. Applied Biosystems does not optimize the data provided by the customer; all rule settings are set by the customer.

Applied Biosystems provides a one-day on-site training with the purchase of the software. This initial training provides setup of the software and some basic tools to start using the software; however, it is not a comprehensive data analysis training program. Applied Biosystems refers to the GeneMapper™ *ID* Software Human Identification Analysis User Guide and Tutorial (2, 3) for reference to the software. Additionally, Applied Biosystems has Technical Support available for telephonic or electronic questions. Furthermore, free webinars (www.appliedbiosystems.com) are available periodically.

Since GeneMapper *ID* is available on the consumer's electrophoresis instrument, it is readily available for analysis and takes virtually no time to run the application. Many features in GeneMapper *ID* are intuitive, most likely since the Implementation Team already has familiarity with Applied Biosystems' software programs. However, for data analysis, GeneMapper *ID* is time-consuming. The software has PQVs (Process Quality Values) for ease in evaluating each sample; however, they are laborious. The hard copy documentation/users reference guide information made available by Applied Biosystems prior to the training is an excellent resource to the Implementation Team.

The analysis of the size standard in GeneMapper *ID* (see Fig. 4) is simple. This analysis tool is effective in efficiently evaluating the size standard.

TrueAllele® System 2

With TrueAllele System 2, the hardware is included with the software installed. The software is not purchased, it is leased throughout the lifetime of its use. The consumer works collaboratively with the staff of Cybergenetics to determine the hardware needs for the system. Upon the delivery of the TrueAllele System 2, the consumer owns the hardware. The hardware for TrueAllele System 2 is simple to assemble. With TrueAllele System 2, due to the optimization process required by this system, Cybergenetics provides a confidentiality agreement. The mutual confidentiality agreement was initiated with the request from the Implementation Team for optimization of the software. Prior to the initial training at Cybergenetics, the optimization was complete for all data provided.

Cybergenetics provides two training sessions, held on site at Cybergenetics, included with the purchase of TrueAllele System 2. The first training session is a 2-day Executive Training provided to the managers and administrators of the laboratory. The Executive Training presents information, from management's perspective, on aspect necessary to integrate the technology into a particular laboratory work process. The training includes an overview of the software system and defines the administrator control features. Following the Executive Training, User Training is provided for the scientists using the software system. The User Training is an intensive 4-day training session using the datasets provided by the consumer for the optimization process. The staff of Cybergenetics helps identify rule thresholds and educates the consumer on the proper navigation of the software while assuring that the scientists understand the utility of the tools available. Additionally, Cybergenetics has Technical Support available for telephonic or electronic questions.

The data for TrueAllele System 2 must be reconfigured prior to being run through the software system. Depending on the number of samples to be analyzed, this partially automated process can take several hours. TrueAllele System 2 provide tools and supporting information that make data review very quick. TrueAllele System 2 was fairly straightforward. The staff of Cybergenetics determined the parameters for the flags and rules during the optimization process and provided explanation during the User Training. The hard copy documentation information made available by Cybergenetics during the training is an excellent resource to the Implementation Team. Cybergenetics requires some *a priori* knowledge of the software before receiving specific documentation.

The analysis of the size standard in TrueAllele System 2 (see Fig. 5) is simple. This analysis tool is effective in efficiently evaluating the size standard.

The Forensic Science Service® DNA Expert System Suite FSS-i³

With FSS-i³, the hardware is purchased separately. Minimum hardware requirements are provided; however, the consumer determines the purchase of the hardware. The laboratory only purchases the software from the vendor. The Forensic Science Service installed the software during the on-site visit to the laboratory. At this time, a fee structure is in place from the Promega Corporation⁴. FSS-i³ requires optimization of the system. The FSS-i³ provided a confidentiality agreement upon the initial request from the Implementation Team for optimization of the software; however, both parties have agreed at this time that a confidentiality agreement is not necessary. Upon training at Marshall University, the optimization process was not complete.

The Forensic Science Service offers either on-site or company-site training. The training program is a separate expense to the purchase of the software. The training program provided in August 2005 by The Forensic Science Service was a five-day installation, optimization and training session. Much of the training time was consumed in an attempt to optimize the analysis of the data. The operation of the software itself is straightforward; keep in mind, however, that the consumer is required to have mastered

⁴ At the time of the presentation, Marshall University and The Forensic Science Service were still discussing and attempting to finalize the fee structure for the software.

either GeneScan®/Genotyper® (Applied Biosystems, Foster City, California) or GeneMapper™ *ID* software prior to using FSS-i³. FSS-i³ builds from the peak detection and sizing algorithms produced from Applied Biosystems' software. The Forensic Science Service has Technical Support available for telephonic or electronic questions⁵.

FSS-i³ builds from data generated with GeneMapper *ID* or GeneScan/Genotyper; GeneMapper *ID* or GeneScan/Genotyper is used to assign base pair size, peak height, and peak area to those peaks detected above the set threshold. These data are imported, in table format, into FSS-i³ where analysis continues. This process requires the manual transfer of data files from these software programs into the FSS-i³ environment. Once the data is imported into the FSS-i³ software, the application run time is minimal. FSS-i³ provides tools and supporting information that make data review straightforward. With FSS-i³, the Implementation Team had difficulty understanding the application and implication on the data of some of the rule options for this software. This, along with technical difficulties related to data file format and data transfers, impacted the ability to complete the optimization process. The Forensic Science Service provided a PowerPoint® presentation at the time of training. No hard copy reference guides were available prior to or during the training. Since FSS-i³ builds upon Applied Biosystems software, the evaluation of the size standard is performed via Applied Biosystems' software rather than post import into FSS-i³.

Conclusion

The objectives of the NEST Project are to evaluate expert systems as a mechanism for the rapid, accurate technical review of convicted offender single-source samples; to hold demonstration/training sessions for the different systems; and to summarize the features and limitations of each software package so that the forensic analyst can make informed decisions on the purchase of such systems. The evaluation of the various software packages is currently underway. Ultimately, expert systems can help reduce the backlog of convicted offender samples and ensure timely submission of these data into CODIS. Individual laboratories will need to closely evaluate and ascertain their throughput needs, management and structure of its agency, budget, human resources, information technology support, and the defined quality assurance/quality control program in order to determine which expert system will most benefit its needs.

The Implementation Team will continue to evaluate all systems and report their progress through this process. It is clear that the Implementation Team has assisted the different vendors by bringing attention to areas where there is opportunity to improve their respective products; the Implementation Team is aware that the vendors are currently working towards improvements on some of the evaluations presented in this paper. The Implementation Team will work with representatives of each vendor for the next five months and are eager to continue the evaluation of their respective expert systems.

Acknowledgments

The authors of this paper would like to acknowledge the Project Team Advisors who continue to contribute to this project: David Coffman, Florida Department of Law Enforcement/SWGDAM Chair; Cecilia Crouse, PhD, Palm Beach Sheriff's Office/SWGDAM Expert Systems Subcommittee Chair; Richard Guerrieri, MS, Federal Bureau of Investigation; John Butler, PhD and David Deuwer, PhD, National Institute of Standards and Technology; Barry Duceman, PhD, New York State Police; Ken Konzak, MS, California Department of Justice; and Tracey Johnson, MSFS, Armed Forces DNA Identification Laboratory.

In addition, we would like to acknowledge the continued support from John Morgan, PhD, Susan Narveson and Lois Tully, PhD of the National Institute of Justice and Deborah Elliotte from Marshall University.

⁵ The responsiveness of inquiries to The Forensic Science Service from Marshall University prior to the collaboration with Promega had been delayed at times, possibly due to the considerable time difference between the two countries.

References

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Table 1.

An expert system for the NEST Project must:
Meet the criteria defined in the National DNA Index System DNA Data Acceptance Standards, Appendix B (1)
Be publicly available for purchase
Be configurable off-the-shelf software
Be completely housed in the purchasing laboratory's facilities
Not require of the user computer code knowledge

Figure 1. Display of a sample in GeneMapper *ID*.

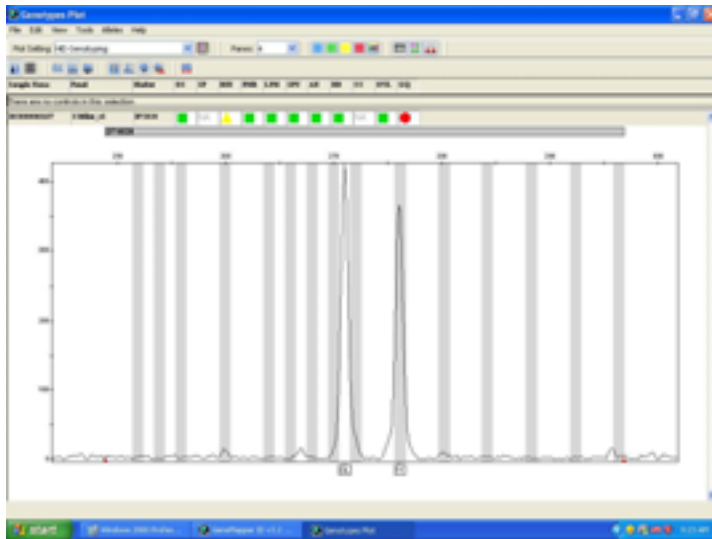


Figure 2. Display of identical sample in TrueAllele System 2 as in Figures 3 and 5.

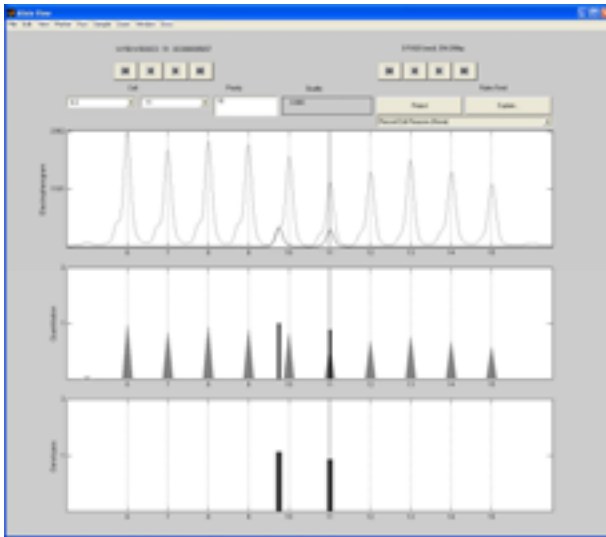


Figure 3. Display of identical sample in FSS-i³ as in Figures 3 and 4.



Figure 4. View of size standard in GeneMapper ID.

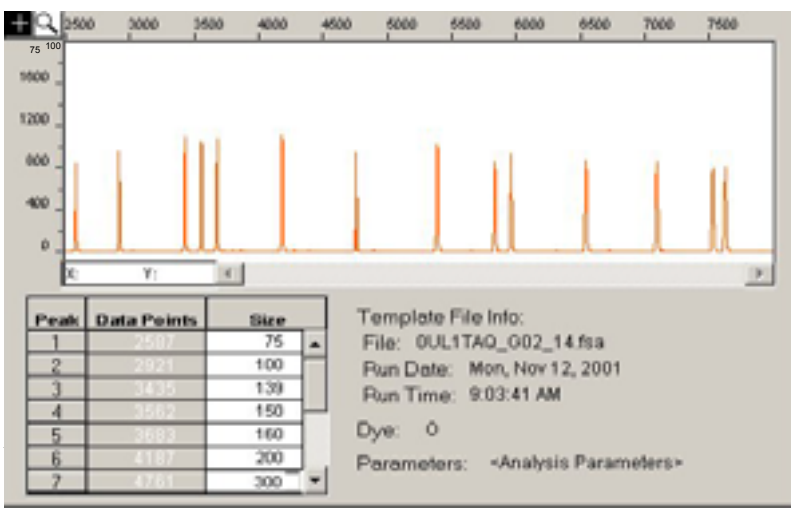


Figure 5. View of size standards for multiple samples in TrueAllele System 2.

