B30   Using Computers to Overcome Forensic DNA Testing Bottlenecks and Improve Recovery From Complex Samples

John Donahue*, Beaufort County Sheriff's Office, 111 Industrial Village Road, Beaufort, SC 29906; and Mark W. Perlin, PhD, MD, Cybergenetics, 160 N Craig Street, Ste 210, Pittsburgh, PA 15213

After attending this presentation, attendees will better understand the potential for automation of DNA mixture interpretation and genotype comparison.

This presentation will impact the forensic science community by demonstrating the advantages of automating DNA mixture interpretation with computer technology.

DNA testing has been utilized in criminal investigations for more than 30 years. In that time, technological advances have improved the ability of laboratories to provide information to investigators and the courts. These gains in technology have primarily come through an improved ability to recover and detect alleles present in the DNA sample, but additional gains have come through automation.

A recent survey of the National DNA Index System (NDIS) -participating laboratories demonstrated that nearly all respondents have implemented automation to some degree. In practice, automation tends to be limited to DNA extraction and setup of DNA quantification, amplification, and sequencing plates. While automation has reduced the time required for completions of these steps, the unintended effect is often a backlog in profile interpretation.

Technological improvements have also increased both the number of samples submitted to laboratories and the complexity of the DNA profiles obtained from these samples. Laboratories are now testing many more sample types than when forensic DNA testing was first implemented. While initial DNA testing was restricted to identifiable body fluids, such as semen, blood, and saliva, improvements have led to testing of various substrates, such as articles of clothing or items that have been handled or manipulated. Such items are often referred to as trace or touch DNA evidence. These samples tend to present mixed profiles showing varying numbers of contributors and mixture ratios.

Mixture interpretation in general can be time consuming, and interpretation of mixtures from trace DNA samples may be very difficult and, at times, too complex for accurate interpretation by human analysts. This complexity has in the past led to many results being reported as inconclusive. Those mixtures that can be interpreted by the human analyst may require a significant amount of time for the analyst to capture all the possible genotype combinations for each contributor and even more time for technical review. This bottleneck leads to an overall increase in case turnaround time.

Contamination assessment is another problem. While some software (e.g., Genemapper® ID-X, the Combined DNA Index System (CODIS)) allows for comparison to reference profiles such as laboratory staff or crime scene investigators, these searches are typically restricted to simple allele-to-allele comparisons. Depending on the number of alleles at each locus, the size of the comparison database, and the search parameters, a simple allele-to-allele comparison can lead to a large number of candidate matches, which must all be reviewed by the human analyst. Conversely, if the search is restricted to reduce the number of adventitious matches, then a contamination event may not be detected.

Problems with mixture complexity, interpretation, and contamination assessment can be alleviated through the use of computer technology to automate DNA mixture interpretation and comparison to reference samples. This presentation will detail the methods that a small laboratory has used to automate DNA profile interpretation with the TrueAllele® computer system.

By using TrueAllele® to automate DNA profile interpretation, a human analyst can upload an entire 96-well plate to the TrueAllele® Investigative Database at the end of the workday, return the next morning, and examine the results. Potential matches and exclusions are identified immediately. In addition, the TrueAllele® Investigative Database can identify matches to every evidence profile and reference sample previously tested, and generate investigative leads to cases already worked.

Automated interpretation may also assist the examiner in the inference of genotypes suitable for export to CODIS. The TrueAllele® software will also automatically compare all mixture data to crime scene investigators and laboratory staff, thus giving the analyst the ability to easily identify contamination. By automating DNA mixture interpretation, the analyst's hands-on time can be reduced, more investigative leads may be identified, more CODIS-eligible profiles may be generated, and all DNA mixtures may be assessed for potential contamination.

DNA Bottlenecks, Computer Automation, Investigative Database