Validation study of the Genophiler® automated data review system

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Abstract

Genophiler® is a software system that automates the processing of GeneScan® and Genotyper®. It also generates summary genotype tables and reports. Genophiler® performs the same actions that an expert user would employ during the course of a review of forensic DNA testing results with GeneScan® and Genotyper®. Genophiler® reports are generated with a simple set of rules that recognize conditions that are indicative of significant peak height imbalance, alleles that are associated with pull-up, and the presence of more than one contributor to a sample. The validation of this system consisted of a review of 25 cases where STR-DNA profiles were generated and comparing the GeneScan® and Genotyper® output to that presented by Genophiler®. Any reported issues (e.g. significant peak height imbalance) flagged by Genophiler® were also manually validated. Full concordance between the output presented by GeneScan® and Genotyper® and Genophiler® was found. Genophiler® is designed to focus the review on the testing results and to eliminate the need for manual software analysis. In addition, we have an ongoing validation process of comparing the results generated by Genophiler® and those produced by the testing lab in every case we review. **This validation study demonstrates that Genophiler® is dependable for the routine evaluation of forensic DNA testing results.**

Genophiler®

- Objective review of DNA evidence
- Flags problems for expert review
- Fast turnaround times
- Organizes all analysis output on a CD-ROM with a simple web interface
- Patent pending

Automation

- Automates all actions to run GeneScan and Genotyper
- All keystrokes and mouse clicks are scripted
- Output saved in PDF format
- Output organized in web page format
- Creates table of alleles and report

Issues Flagged

- Potential mixtures
  - Observing 3 or more alleles at any locus
- Peak height imbalance
  - Observing heterozygous loci with peaks differing by more than 30% (threshold can be adjusted)
- Pull-up
  - Observing two peaks in different dyes at approximately the same position (± 0.04 bp)
  - All observed instances were of the exact same size

Table of Alleles

- Samples organized by type
- Color-coded alleles
  - Alleles consistent with victim are red
  - Alleles consistent with defendant are blue
  - Alleles consistent with victim and defendant are green
  - Alleles not consistent with victim or defendant are black

Genophiler® Summary Tables

Pull-Up

Peak Height Imbalance

Mixture

Genophiler DNA Analysis System

Genophiler® Report

Validation

- 25 cases examined
- 740 profiles in the allele tables were consistent with their electropherograms
- 124 instances of pull-up (in 87 samples) were consistent with pull-up (sizes differing by less than 0.04 bp)
- 169 instances of peak height imbalance (in 129 samples) contained heterozygous loci with peaks differing by more than 70%
- 389 samples were potential mixtures (having loci containing three or more alleles)

Conclusions

- Genophiler® accurately stores and presents the data found in Genotyper® electropherograms
- Genophiler® accurately identifies the issues of peak height imbalance, pull-up, and potential mixtures
- Genophiler® is a reliable tool for assisting in the evaluation of DNA evidence

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