B212 An Analysis of an Internal Validation Dataset for the New Core Short Tandem Repeat (STR) Loci

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After attending this presentation, attendees will better understand the open source interpretation tools for analyzing large internal validation data sets.

This presentation will impact the forensic science community by exploring different means to parse the output from internal validation experiments designed and performed in forensic laboratories to demonstrate the reliability, reproducibility, and robustness of the current larger STR DNA genotyping chemistries.

In March of 2015, the Federal Bureau of Investigation (FBI) published the expansion of the original core 13 loci to a new Combined DNA Index System (CODIS) core containing 20 loci.[1] Following validation guidelines outlined by the Scientific Working Group on DNA Analysis Methods (SWGDAM) and the European Network of Forensic Science Institutes (ENFSI) DNA Working Group, an internal validation dataset was generated to assess the PowerPlex® Fusion™ 6C STR multiplex chemistry.[2,3]

Organization and analysis of internal validation data is often performed manually within an Excel® spreadsheet format within laboratories. Thorough analysis of validation data sets for the new STR typing kits and/or new technologies within forensic laboratories may appear daunting, but it is essential for robust data interpretation to generate the most accurate results for producing a laboratory's standard operating procedures and technical manuals.

To illustrate the analysis of an internal validation data set, 44 reference and known samples were used in experiments designed to validate PowerPlex® Fusion™ 6C, which is comprised of 23 autosomal STRs, 3 Y-chromosomal Short Tandem Repeats (Y-STRs), and amelogenin. Results obtained from the internal validation experiments provided data to extract and evaluate parameters, such as sensitivity, stochastic effects, sizing precision, allele calling accuracy, repeatability and reproducibility, DNA mixture performance, and contamination detection.

This presentation will describe the process of interpreting internal validation experiments for PowerPlex® Fusion™ 6C using several open source software tools. Data analysis were conducted using the in-house software programs developed at the National Institute of Standards and Technology (NIST) (http://www.cstl.nist.gov/strbase/software.htm), as well as *STR-validator*, a software created by Oskar Hanson at the Norwegian Institute of Public Health.[4]

This presentation will illustrate: (1) the specific data formatting for import into software tools; (2) the calculations of analytical and stochastic thresholds manually as well as in *STR-validator*, which examines multiple published calculation methods; and, (3) the data output of stutter percentage calculations, peak height ratios, base-pair sizing precision, mixture detection, genotyping concordance, reproducibility, and sensitivity.[5-7] The goal is to share the findings of this study with forensic laboratories and introduce the community to open source programs that can be utilized during the internal validation process.

Reference(s):

1. Hares D.R. Selection and implementation of expanded CODIS core loci in the United States. *Forensic Sci Int Genet.* 2015. 17: p. 33-4.

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2. SWGDAM Validation Guidelines. Available from: http://www.az-forensics.com/docs/pdfs/SWGDAM_Validation_Guidelines_APPROVED_Dec_2012.pdf.
3. ENFSI. Recommended Minimum Criteria for the Validation of Various Aspects of the DNA Profiling Process.pdf.
4. Hansson O., Gill P., Egeland T. STR-validator: an open source platform for validation and process control. *Forensic Sci Int Genet.* 2014. 13: p. 154-66.
5. Gill P. et al. Genotyping and interpretation of STR-DNA: Low-template, mixtures and database matches-Twenty years of research and development. *Forensic Sci Int Genet.* 2015. 18: p. 100-17.
6. Monich U.J. et al. Probabilistic characterisation of baseline noise in STR profiles. *Forensic Sci Int Genet.* 2015. 19: p. 107-22.
7. Bregu J. et al. Analytical thresholds and sensitivity: establishing RFU thresholds for forensic DNA analysis. *J Forensic Sci.* 2013. 58(1): p. 120-9.

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