VALIDATION OF GENEMAPPER® ID-X V1.1.1 INCLUDING A TIME AND COST-BASED COMPARISON STUDY WITH GENEMAPPER® V3.2.1

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Abstract

With the bottleneck of forensic DNA analysis moving from sample processing to data analysis, affective computer software is needed to decrease the amount of time and resources spent analyzing samples. GeneMapper® *ID-X* v1.1.1 (GMIDX) was validated for use as a partial expert system, utilizing the allelic ladder and control assessment tool for forensic DNA data analysis. A comparison of this software with GeneMapper® ID v3.2.1 (GMID) and a time and cost evaluation were completed. The allelic ladders, controls, and size standard assessment tools performed reliably and will be implemented. Identical results were obtained when analyzed using both programs. GMIDX allowed for more efficient data review. The Profile Comparison and Mixture Tool will be an aid to analysts.

Introduction

The processing of samples from accession to data analysis requires a significant amount of time and resources. With many forensic laboratories automating sample processing, the bottleneck has moved toward data analysis. For high-throughput laboratories, approximately 50% of their resources can be spent on data analysis.[1]

An expert system is the collaboration of known experts in a particular field with computer programmers.[2] The first computer software systems were designed to change the signal from the instrument into a genotype call.[3] Now, expert systems can analyze all possible circumstances that can be encountered and respond accordingly.[4] Because they provide an explanation for the decisions made, these systems allow analysts to review large volumes of forensic samples quickly.[5]

GeneMapper® *ID-X* v1.1.1 was designed to help streamline and maximize the efficiency of data analysis and reduce the amount of time that analysts spend looking at data. As an expert system, GMIDX v1.1.1can automatically review allelic ladders, samples, and controls using user and software defined thresholds. The software can also provide detailed information in the form of color-coded Process Quality Value flags (PQV), which can help in the manual review of samples.[6]

Materials

1. Applied Biosystems (AB) GeneMapper® *ID-X* v1.1.1 Software
2. AB GeneMapper® *ID* v3.2.1 software
3. .fsa data generated by an AB Genetic Analyzer 3130, using Promega Corporation's PowerPlex®16 HS, AB Identifiler®, and AB Yfiler® amplification kits

Methods

Allele calls, base pairs, and height in relative fluorescence units (RFU) for each sample were compared to the GeneMapper® ID v3.2.1 produced data. Artifact calls were compared and were changed using the "rename allele" feature in GMIDX v1.1.1. All sample PQV flags in GMIDX v1.1.1 were recorded.

The mixture tool was evaluated by the analysts through the use of hand deconvolution, and then the aid of the GMIDX v1.1.1 mixture tool. Results were compared to those in the selected genotypes table within the mixture tool.

A profile comparison tool study was performed for use as a contamination and sample concordance check. The percent match threshold was set to both 70% and 80%. DNA profiles for 41 laboratory employees were uploaded into the profile comparison tool and compared to a data project containing three instances of contamination by laboratory personnel.

Five previously analyzed samples with a variety of typically encountered artifacts were evaluated. Allele calls were edited and re-edited and the allele history was analyzed and documented.

Additionally, a time and cost savings study was performed by evaluating the time required to review allelic ladder and control profiles for both the first and technical review. This value was used to determine the total time and cost saved based on the projected number of cases that would be evaluated in 2010 and an average of the laboratory's analysts' salaries.

Table 1: Sample Totals

	Identifiler®	Yfiler®	PowerPlex® 16 HS	Total
Ladders	31	48	39	118
Positive Controls	7	20	28	55
Negative Controls	20	34	20	74
Samples	33	32	55	120
Total				367

Table 2: Artifacts Indicated by GMIDX v1.1.1

Kit	-A	+A	PU	TA	OL	ST	RB	DO	PP16	WA	Total
Identifiler®	1	0	5	2	24	4	2	15	0	4	57
Yfiler®	0	1	12	0	1	20	7	0	0	0	41
PowerPlex® 16HS	0	2	39	0	52	29	26	31	15	0	197
Total	4	3	56	2	77	53	35	46	15	4	295

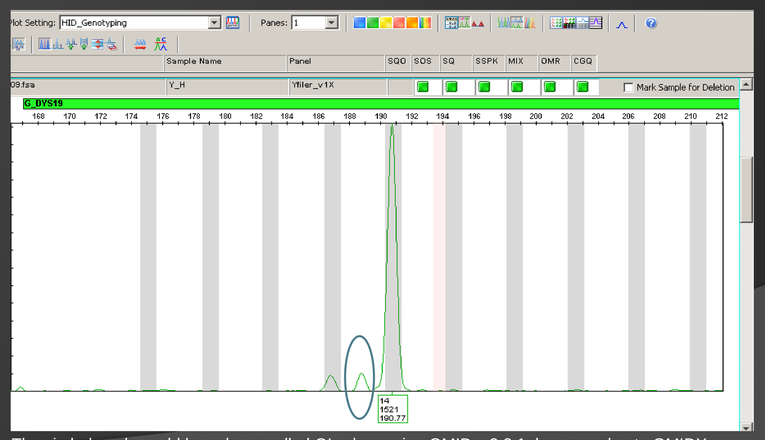
Key: -A- Minus A, +A- Plus A, PU- Spectral Pull Up, TA- Triallele, OL: Off Ladder, ST- Stutter, RB- Raised Baseline, DO- Drop Out, WA- Wrong Allele Call, PP16- PowerPlex 16 HS known artifact

Table 3: Flags Indicated by GMIDX v1.1.1

Kit	AN	PHR	LPH	CC	MPH	BIN	GQ	OMR	BD	SPK	OS	Total
ID	31	21	11	22	7	18	55	2	0	0	0	167
YF	15	30	67	5	7	11	133	3	1	0	0	272
PP 16HS	75	120	94	29	37	36	308	2	1	13	50	765
Total	121	171	172	56	51	65	496	7	2	13	50	1204

Key: AN- Allele Number, PHR: Peak Height Ratio, LPH- Low Peak Height, CC- Control Concordance, MPH- Maximum Peak Height, BIN- Out Of Bin, GQ- Genotype Quality, OMR- Out Of Marker Range, BD- Broad Peak, SPK- Spike, OS- Off Scale

Figure 1: Stutter at DYS19 with GMIDX v1.1.1



The circled peak would have been called OL when using GMID v 3.2.1, however due to GMIDX v1.1.1's multiple stutter filters, a designation was not given.

Results

- GeneMapper® *ID-X* v1.1.1 correctly analyzed 247 allelic ladders, positive controls and negative controls as seen in Table 1: Sample Totals
- Allele calls, sizing calls, and RFU values were concordant between the software systems.
- A total of 295 artifacts were detected using the GMIDX v1.1.1software as noted in Table 2: Artifacts Indicated by GMIDX v1.1.1.
- Artifacts located between markers are not labeled in GeneMapper® ID v3.2.1, but are labeled in the GMIDX v1.1.1 software.
- GMIDX v1.1.1 did not always correctly label artifacts, for example, a spectral pull-up peak was incorrectly labeled as a spike.
- Some YFiler® samples contained off ladder alleles in GMID v3.2.1, but were filtered out as stutter in GMIDX v1.1.1, as illustrated in Figure 1: Stutter at DYS19 with GMIDX v1.1.1.
- Neither software could detect allelic dropout.
- The analysts found the mixture tool useful, however, it was not useful for mixtures containing low level minor contributors.
- The profile comparison tool was effective at determining similarities among samples and employee profiles. The 80% Match Threshold was chosen because it yielded the most informative number of matches.
- The allele edit audit documents were reviewed and found to correctly record all allele edits made.
- The use of GMIDX v1.1.1 as a partial expert system will save an estimated 60 minutes of analysis time per project. Based on the 382 case projection for 2010, assuming five samples per case, the Washoe County Sheriff's Office will be saving approximately 382 hours of data analysis and review time.
- A total of approximately $12,532-15,923 will be saved by analyzing data with GMIDX v1.1.1, a projection that includes both salary and the cost of supplies.

Conclusions

- GMIDX v1.1.1 is an effective means of reducing time and resources, without sacrificing quality, when analyzing single source samples due to the implementation of allelic ladder, control, and size standard quality assessment tools.
- The Analysis Summary Window was proven to be useful in organizing samples according to their type and possible anomaly.
- GMIDX v1.1.1 was capable of alerting the analyst to samples that may require further processing by using color-coded PQV flags, as seen in Table 3: Flags Indicated by GMIDX v1.1.1.
- While the mixture and profile comparison tools will not be used extensively, they will be a welcome addition to the data analysis workflow.
- The Washoe County Sheriff's Office will need to prepare new protocols for the analysis of single source samples utilizing the GMIDX v1.1.1 quality assessment tools.
- Based on the results of this validation, the Washoe County Sheriff's Office plan to test the use of GMID-X v1.1.1 as an expert system for first review of CODIS (Combined DNA Index System) samples.

References

1. Harbison, A., McCabe, B. Power, T. FaSTR DNA: A new expert system for forensic DNA Analysis. Forensic Sci Int 2008; 159-165

2. Lynch, KJ, Rodgers, FJ. Development of integrated criminal justice expert systems applications. Artificial Intelligence Laboratory Management Information Systems; 13 Aug 2002. Tuscon: Eller College of Management The University of Arizona http://ai.eller.arizona.edu/COPLINK/publications/develop/developm.html.

3. Butler, J. M. Forensic DNA Typing, 2nd Edition, 2005.

4. Roby, R. Expert systems help laboratorys process DNA samples. NIJ Journal 2008; 16-19

5. Roby RK, Jones JP. Evaluating expert systems for forensic DNA laboratories. Forensic News. Foster City: Applied Biosystems, 2005 http://docs.appliedbiosystems.com/pebiodocs/00115112.pdf.

6. Applied Biosystems. Genemapper® *ID-X* v 1.0 Product Bulletin. Foster City, CA: Applied Biosystems. 2007.

Acknowledgements

I would like to thanks Dr. Lisa Smyth-Roam, Washoe County Sheriff's Office Technical Leader, Jason Chute, Marshall University Forensic Science Center Technical Leader, and Dr Pamela Staton, Marshall University topic advisor for all of their help and support. I would also like to thank Justin Godby for preparing me for this project, and assisting with the creation of this poster. This project was supported by Award No. 2005-MU-BXK020 awarded by the National Institute of Justice, Office of Justice Programs, U.S. Departments of Justice. The opinions, findings, and conclusions or recommendations expressed in this publication/exhibition are those of the author(s) and do not necessarily reflect the views of the Department of Justice.