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Work in "progress"—Applied Biosystems GeneMapperID™

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Abstract. Higher sample throughput in most forensics labs comes along with an increasing need of suitable automated fragment analysis and allele calling software. Applied Biosystems' GeneMapperID™ was introduced to replace time consuming softwares GeneScan® and/or Genotyper® including an Oracle™ database and multi-user functionality. The present description shows that the GeneMapperID™ offers a number of improvements in handling and viewing, but also exhibits a wide range of bugs and annoying alerts up to the danger of wrong allele calling. Embedded in the 'Integrated Solutions' philosophy of AB, which makes labs more and more dependent on AB products, a critical appraisal is taken on the GeneMapperID™ and its "work in progress" characteristics.

Keywords: GeneMapperID™; Bug; HID testing

1. Introduction

From the user guide to GeneMapperID™ 3.1: "...The GeneMapperID™ Software Version 3.1 combines functions of the GeneScan® and Genotyper® Software...is designed specifically for AmpFlSTR data analysis...has not undergone specific developmental validation for human identification..."

The GeneMapperID™ software solution for forensic identity testing is finding its way into more and more laboratories worldwide. Replacing the time consuming GeneScan® and/or Genotyper® softwares, the user expected to purchase a modern, easy-to-use software that performs high quality genotyping and provides a wide range of options in the field of forensic DNA testing.

Our Institute uses the GeneMapperID™ frequently for the analysis of commercial STR-multiplexes. In comparison to GeneScan® and Genotyper® softwares, one must admit that

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the combination of these two softwares succeeds in all basic functions. The new software is faster, easier to handle, better plot views are possible and a lot of functions to improve the genotyping quality were added, at least compared to GeneScan®/Genotyper®.

Compared to other software solutions, e.g. the 'ancient' ALFwin™ by Amersham Biosciences, most of the functions that were presented as 'new', especially handling functions for viewing, zooming and plotting, tables were already available in 1995.

The present description gives an overview on the experiences made with the GeneMapperID™ throughout the different stages of application, beginning with the system requirements/installation and ending with a critical appraisal of AB's policy concerning the ongoing restrictions through so-called 'integrated solutions'.

2. System requirements/installation

From the release note attached to the update to Version 3.2: "Please do not select items from a dropdown list using keyboard commands. . . . Select items by using mouse pointer."

- Recommended hardware (PIII, 700 MHz, 512 MB RAM): We are using the software on a Pentium IV with 3.0 GHz, 1 GB RAM and a high end graphics card. Even with this system, the software runs sometimes low.
- Installation takes long, probably because of the Oracle database, but finally succeeded. Possible reinstallations might be necessary.
- First step/first problem: Runfiles and allelic ladders have to be imported from a single run folder. Not mentioned in the "Fast Guide", but after all once in the user guide: "Considerations for HID analysis." It took some time to find it.

3. Experiences, problems, bugs and own faults

From the user guide to GeneMapperID 3.1.: "Troubleshooting sheets: To address any issues that may arise when installing or running the software, please fill out the appropriate list of questions, and collect the necessary files before contacting technical support." From the nice women at AB, Darmstadt: "I am not able to connect you to the technical support when you use this number. Please call. . ."

- Allelic ladders can only be viewed dye per dye. A combination (possible only with the overlay function) of all dyes in four separate panels is impossible.
- The Size Match Editor window opens in a small-sized window. Checking the internal lane standard properly requires stretching of the window or zooming. Every time. Uncomfortable.
- The new Software Version 3.2 provides the option to display label peak assignments for the size standard as it was already possible in Genotyper®. Editing of the size standard in this view is not possible. Absurd.
- After updating from Versions 3.1 to 3.2: The display font size of the peak labels is ignored when printing. The fonts/labels are bigger as determined in the display view. This is no printer problem. Possible solution: reinstallation, buy more paper.
- Switching from one color to the other in the Sample Plot View or closing Sample Plot Windows takes 1–2 s. Too long for simple graphics with peaks.

- Alert: More than one peak will be deleted!: What a surprise after selecting several peaks to delete them. This alert is not necessary, but annoying.
- Even with bad quality data, GMID™ assigns the peaks of the allelic ladder. This leads to wrong typing results. Mistakes of the user: peak thresholds too low, bad spectral calibration/matrix, bad capillary. Here an alert would be necessary, not only a small quality flag in the genotype table. Or better: Bad quality ladders could be refused by GMID™ for analysis.
- Alert "Unable to save Panel Data...!": Importing Panels and Bins, especially from other STR-Kit-manufacturers is a severe problem for GMID™. According to AB support, this is a function to protect Bins and Panels that are already imported against damage. Believe it or not. Reinstallation is useless.
- Exporting more than one project in a row is possible. But for every project a new file name has to be defined. It is not possible to export the projects fast under the original name. For reinstallation and backup purposes, this would be nice. For analysis methods, table and plot settings and size standards, every setting has to be exported separately. This takes a lot of time.

4. 'Integrated solutions'

Throughout the last years, AB products for human identification and forensic applications get more and more specialized to particular AB instruments, chemistry or software. The user is forced to spend a lot of time to adopt previously used protocols, matrices and run methods on their machines for use with the newer software versions and technical solutions, such as new Matrix Standards, Data collection or GeneMapperID® Versions 3.1 and 3.2. Sometimes, methods can no longer be used. New methods or kits (e.g. SNPlex™) can only be used after upgrading the instrument or buying newer versions of special software. Software upgrades like from Data collection 1.0 to 2.0 force the user to abandon functions and options that were useful in the earlier version of the software. Expensive additional maintenance is necessary to get the same result as before. In some cases, "integration" appears to offer predefined solutions, which complicate or even prevent user-defined control of the sample processing.

5. Conclusion

As AB products benefit from the experiences made by the users since many years, one could expect an ongoing interest in cooperation. The current way AB deals with its customers does not acknowledge this adequately. As many users are confronted with these developments, a critical discussion is necessary.