



B72 GeneMapper™ ID Software Test Plan Performed for Software Verification

Rhonda K. Roby, MPH*, Cherisse Boland, BS, Yasser Daoudi, BS, Liwei Qi, MS, and Brenda Takara, MS;
Applied Biosystems, 850 Lincoln Centre Drive, M/S 404-3, Foster City, CA 94404

The goal of this presentation is to summarize the verification performed for GeneMapper™ ID Software v. 3.1, an integrated fragment analysis software application, on databasing and forensic-simulated samples amplified with AmpFISTR® PCR Amplification Kits.

This presentation will describe and verify an excellent tool for genotyping of samples for forensic, databasing, and parentage applications.

GeneMapper™ ID Software v. 3.1 (Applied Biosystems, Foster City, CA, USA) was specifically designed to be a complete integrated software solution for forensic, paternity, and databasing laboratories performing STR analysis using ABI PRISM® genetic analysis instruments and AmpFISTR® PCR Amplification Kits for automated genotyping. This software analyzes the raw data collected from the ABI PRISM® instrument platforms and automatically identifies peaks, quantifies signal intensity, sizes each DNA fragment, and makes allele calls using defined panels and bin sets with its automated genotyping capabilities. A test plan, which Applied Biosystems has defined as verification of the software, was designed to develop a set of test criteria to evaluate the performance of this software in the human identification communities. The test plan lists approximately 50 key elements to evaluate the software's robustness, performance, and feature design.

Leibelt, *et al.* first introduced GeneMapper™ ID Software v. 3.1 at the American Academy of Forensic Sciences Annual Meeting, 2003. Additionally, a comprehensive concordance study of STR profiles of forensic-simulated samples (e.g., mixtures, degraded DNA, and inhibited DNA) generated with six (6) different AmpFISTR® PCR Amplification kits (Identifiler®, Profiler Plus®, COfiler®, SGM Plus®, and SEfiler™ kits), analyzed on four (4) ABI PRISM® genetic analysis instruments, and using various ABI PRISM® software packages for both data collection and data analysis was presented by Boland *et al.* (American Academy of Forensic Sciences Annual Meeting, 2003). Samples were processed on the ABI PRISM® 310 Genetic Analyzer (for both Macintosh® and Windows NT® operating systems), 377 DNA Sequencer (for both Macintosh® and Windows NT® operating systems), and 3100 Genetic Analyzer and 3100-Avant Genetic Analyzer. All samples were then analyzed using both GeneScan® software and Genotyper® software and compared to GeneMapper™ ID Software v. 3.1. Data were analyzed with GeneScan® Software v. 3.7.1 and Genotyper® Software v. 3.7, for use with Windows NT® OS; GeneScan® Software v. 3.1.2 and Genotyper® Software v. 2.5.2, for use with Macintosh® OS; and GeneMapper™ ID Software v. 3.1 in Classic and Advanced modes. The combination of these studies and associated data have led to a comprehensive verification test plan for GeneMapper™ ID Software v. 3.1.

The verification test plan requires that features such as workflow; data handling; peak detection and sizing; HID algorithm testing; and CODIS functions be evaluated with a pass/fail criteria. These criteria include: allele number error for loci containing more alleles than specified in the analysis method; out of bin alleles; peak height ratios, low peak height and spectral pull-up levels specified by the user; broad peaks when the width of the called peak is wider than a specified value; offscale; control concordance when the designated control sample's genotype does not exactly match the definition; and overlap for peaks positioned within the overlapping size range of two markers.

GeneMapper™ ID software includes three peak detector algorithms allowing different levels of user control over data analysis. The "classic" mode produces very similar results as those analyzed with GeneScan® Software v. 3.1.2 designed for the Macintosh® operating system. This algorithm aids in the adoption of GeneMapper™ ID software for laboratories currently using GeneScan® software developed for use with the Macintosh® OS wanting to maintain current interpretation guidelines. The "advanced" mode provides the user with the same analysis parameters available in GeneScan® Software v. 3.7.1, designed for use with the Windows NT® operating system, including several improvements made to the algorithm. An additional "basic" mode allows for analysis using limited parameters consisting of a user defined minimum peak height threshold. Additional features new to the software include CODIS export functionality, automated sample concordance checking and search capability within the GeneMapper™ ID software database. The design for verification testing and the results from the verification testing will be presented.

ABI PRISM and its Design, AmpFISTR, Applied Biosystems, COfiler, GeneScan, Genotyper, Identifiler, Profiler Plus, and SGM Plus are registered trademarks of Applied Biosystems Corporation or its subsidiaries in the U.S. and certain other countries. AB (Design), Applied Biosystems, GeneMapper, and SEfiler are trademarks of Applied Biosystems Corporation or its subsidiaries in the U.S. and certain other countries. Mac and Macintosh are registered trademarks of Apple Computer, Inc. Windows NT is a registered trademark of the Microsoft Corporation. All other trademarks are the sole property of their respective owners.

Software Verification, GeneMapper™ ID Software v. 3.1, AmpFISTR® PCR Amplification Kits