B58 Verification of GeneMapper™ *ID* Software for STR Analysis

Craig S. Leibelt, MS, Cherisse E. Boland, BS, Candia L. Brown, BS, Tony L. Hatch, BS, Yasser Daoudi, BS, Yuandan Lou, PhD, and Rhonda K. Roby, MPH, Applied Biosystems, 850 Lincoln Centre Drive, Foster City, CA*

The goals of this research project are to present an integrated fragment analysis software application for STR analysis and to describe the verification performed and results obtained.

ABI PRISM® GeneMapper™ *ID* software (Applied Biosystems, Foster City, CA) provides automated genotyping for linkage analysis, SNP validation, population genetics and other DNA fragment analysis applications. GeneMapper™ *ID* software was specifically designed to be a complete solution for forensic laboratories performing STR analysis using ABI PRISM® genetic analysis instruments and AmpFlSTR® PCR Amplification kits. Currently, many forensic laboratories are using GeneScan® and Genotyper® software to perform analysis and genotyping of STR profiles. GeneScan® software analyzes the raw data collected from the ABI PRISM® instrument platforms and automatically identifies peaks, quantitates signal intensity, and sizes each DNA fragment. After the data analysis step with GeneScan® software, sample files are imported into Genotyper® software. Genotyper® software is then used for the automated genotyping of alleles when used with specific Genotyper® software template files designed for use with the AmpFlSTR® kits. GeneMapper™ *ID* software integrates the major functions of GeneScan® and Genotyper® software within one application and provides additional capabilities to aid the user.

GeneMapper™ *ID* software provides a graphical user interface which combines a sizing view with a genotyping view to allow for analysis and editing of alleles within the same window. The user interface also provides for the ability to observe samples on a per marker basis or on a whole sample basis. All views pertinent to forensic analysis are supported with GeneMapper™ *ID* software, including the ability to view raw data and analyzed data with colors displayed independently or overlapped. High throughput analysis is addressed with the incorporation of process component-based quality values (PQV), which monitor major components of the sizeand allele-calling process. The quality values are reported by GeneMapper™ *ID* software as an aid to flag criteria related to sample preparation, PCR, separation, detection and analysis on a per marker basis. These quality values are represented as symbols to reflect “pass,” “check,” or “low quality” and are weighted by the user.

The PQV criteria related to genotypes are Boolean in that the sample either passes or fails a specific criterion. These criteria include: allele number error for markers containing more alleles than specified in the analysis method; out of bin alleles; peak height ratios, low peak height, and spectral pull-up levels below that specified by the user; broad peaks when the width of the called alleles’ 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. This includes the “classic” mode which generates the same results as those generated from GeneScan® software version 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 wishing to maintain current interpretation guidelines while upgrading to an improved software application. The “advanced” mode provides the user with the same analysis parameters available in GeneScan® software version
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.

An extensive study to verify genotype concordance and the overall robustness of the features within GeneMapper™ *ID* software was performed. Here, six (6) AmpFlSTR® PCR Amplification kits including Profiler Plus™, Profiler Plus™ *ID*, COfiler®, SGM Plus®, Identifile®, and SEfiler™ kits were used to amplify a set of forensic type samples. The samples were then run on four instrument platforms including the ABI PRISM® 377 DNA Sequencer (for use with both Macintosh® OS and Windows NT® OS), 310 Genetic Analyzer (for use with both Macintosh® OS and Windows NT® OS), and 3100 Genetic Analyzer. This paper will present the results of this study as well as illustrate the features present in GeneMapper™ *ID* software.

GeneMapper™ *ID* Software, Automated Genotyping, AmpFlSTR® PCR Amplification Kits

 ** Presenting Author*