GeneMapper® *ID-X* Software v1.1: Development of an Automated Approach for the Deconvolution of DNA Mixtures

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Crime laboratories are increasingly adopting automated systems as a means of increasing sample processing throughput. As a result of successful implementation of these high-throughput systems, laboratories are faced with the task of reviewing a growing amount of sample data, effectively moving the bottleneck downstream. Casework analysts spend the majority of their hands-on time in two areas, a) the screening and documentation of items containing biological evidence and b) the analysis of complex electronic data to maximize information recovery from DNA samples, especially those containing mixtures. The interpretation of DNA mixtures presents particular challenges for the casework analyst. Significant variation exists in the procedures for evaluating DNA mixtures between laboratories, as they are not standardized, resulting in potentially different interpretations of the same data. In addition, analysis is complicated by factors inherent to fragment analysis which may impact a mixed DNA profile such as: allele dropout due to low input amounts of sample DNA, stutter, artifacts, and heterozygote peak balance at various input amounts.

The GeneMapper® *ID-X* mixture analysis tool is designed to streamline the interpretation of profiles from mixed DNA samples by automating functions which would otherwise be performed manually. The mixture analysis tool assists the casework analyst in performing:

a) Segregation of samples based on the minimum number of contributors
b) Deconvolution of 2-person mixtures into contributor genotypes
c) Statistical analysis of samples incorporating a variety of approaches

An approach to the deconvolution of 2-person mixtures into individual profiles, also referred to as the major and minor contributor, will be described. The approach leverages two key inferences: (1) At any locus, two alleles originating from the same person have roughly the same height, and (2) Established mixture proportions remain consistent across all loci within a sample profile. The approach results in a set of genotype combinations that are scored based on consistency of the mixture proportion at all loci across the profile and heterozygote peak height ratio (measured against user-defined thresholds). An “Inclusion Quality” Process Component-based Quality Value (PQV) is generated which provides an assessment and ranking of the set of genotype combinations. The genotype combinations are segregated into 2 tables based on passing or non-passing Inclusion Quality values. All genotype combinations are available for review by the analyst within the sample plot, allowing analysts to verify the mixture interpretation and make changes based on their knowledge and experience. The software then utilizes the resulting profiles to compute Random Match Probability (RMP), Combined Probability of Inclusion/Exclusion (CPI/CPE) and Likelihood Ratio (LR) statistics commonly utilized by forensic practitioners to provide a measure of the weight of the evidence. All information generated by the tool can be exported for record keeping.