

VALIDATION OF THE MIXTURE ANALYSIS TOOL IN GENEMAPPER® *ID-X* V1.1 SOFTWARE

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More than 50% of the evidence DNA samples submitted to crime labs are mixture samples. Interpreting mixture DNA samples has been a big challenge for crime labs due to the facts that manual interpretation is not only time consuming but also often gives inconsistent results based on different approach and assumption made by each analyst. The integrated mixture analysis tool in the GeneMapper® *ID-X* v1.1 software is designed to automate and streamline the interpretation of DNA profiles from mixed samples and assist the casework analyst to quickly:

- Segregatesamples based on the minimum number of contributors
- Deconvolute of 2-person mixtures into contributor genotypes
- Generate Accurate statistical analysis of sample data

The GeneMapper® *ID-X* v1.1 Software Validation Study was performed to evaluate software's capability in interpreting DNA mixtures by analyzing a set of data consisting of single source and mixed samples amplified with the AmpF ℓ STR® Identifiler®, Profiler Plus®, COfiler®, and SGM Plus® kits generated on the 3130xl Genetic Analyzer. Different mixture ratios and DNA input amounts were prepared for evaluation. The calculations used by sample segregation, pattern deconvolution, and statistical analysis algorithms were compared to the results generated from validated third party scripts. In additional, this validation study also verified the software functionality, workflow, user interface, and documentation.

The test results shown that the GeneMapper® *ID-X* v1.1 software supports data from all the tested AmpF ℓ STR® kits with user-friendly workflows, functionalities, and interfaces for data input, sample pruning and segregation, pattern extraction, known matching, multiple kits merging, and statistics for random match probability, combined probability of inclusion/exclusion, and Likelihood Ratio. The mixture analysis tool also provided accurate calculations for minor contributor mixture proportion (Mx), average Mx and residual, correctly filtered peak height ratios based on user settings, and generated accurate flags for peak height ratio threshold, residual threshold and inclusion quality.

Overall, the GeneMapper® *ID-X* v1.1 software met the design requirements for analyzing mixture samples with 1, 2, and 3 or more contributors. Data from known mixtures demonstrated that the software effectively functions as an automated tool to deconvolute samples into the most likely major and minor contributors with or without known matching. The software correctly merged the deconvoluted profiles of data generated from replicate samples amplified with multiple AmpF ℓ STR® kits. The software also provided correct statistical calculations using defined population databases. The results of the automated mixture analysis were found to be affected by data quality and the analysis parameter settings in that low level peaks exhibited more variable results. In conclusion, by providing consistent mixture interpretation functionality, GeneMapper® *ID-X* software v1.1 mixture analysis tool helps forensic analysts to minimize the forensic data analysis bottleneck and make key interpretation decision more quickly with better confidence.