Human identification

GeneMapper PG Software v1.0

Thermo Fisher

Forensic DNA mixture interpretation software

Introduction

In today's rapidly evolving forensic landscape, the ability to extract meaningful conclusions from complex DNA mixtures is more crucial than ever. Traditional methods often fall short in addressing the challenges posed by crime scene samples. Now, a new chapter begins: an innovative approach to probabilistic genotyping that mirrors the way you traditionally interpret DNA evidence.

Enter Applied Biosystems[™] GeneMapper[™] PG Software v1.0. This advanced tool offers fast, flexible models and clear, real-time explanations and visualizations, moving beyond black-box and overly complex methods. GeneMapper PG Software offers clarity without compromise. The software's transparent, scientifically rigorous model and robust algorithms are designed to align with the thought process of a forensic analyst. Built on decades of real-world experience, even challenging mixtures are handled with speed, reliability, and courtroomready confidence.

Move from complexity to clarity

GeneMapper *PG* Software can assess an entire project of evidence and reference samples to identify potential contributors to mixed-source samples. This powerful tool helps answer critical questions such as:

- Is a person of interest (POI) part of this mixture?
- How does the likelihood ratio (LR) change if the POI is replaced by an unknown–unrelated or related contributor?
- What are the individual deconvoluted profiles in an evidence sample?
- How many contributors does this sample have?
- Which evidence samples contain potential contributors?
- Which potential contributors are promising leads?
- What do two samples have in common (i.e., do they contain a common POI)?

Utilizing the deterministic biological context optimized sampling (BCOS) algorithm that closely mirrors manual analytical processes, GeneMapper *PG* Software can support enhanced reproducibility and interpretability. Complex formulas and calculations are accessible, with the user interface displaying the logic in real time. This enables consistency, as results remain the same with repeat analysis. Powerful visualization tools help explain results and allow detailed analysis exploration. GeneMapper *PG* Software enhances forensic analysis by facilitating clear, insightful answers, enabling informed decisions with confidence.

GeneMapper PG Software v1.0 features

- **Compare profiles:** Compare profiles across your project, whether evidence-to-evidence or evidence-to-references
- Number of contributors (NoC) estimation: Utilizes multiple algorithms for contributor estimation, including:
 - Decision tree analysis
 - Total peak analysis
 - Maximum allele count (MAC) analysis
- **Deconvolution:** A fully continuous, probabilistic model for reproducible mixture deconvolution including:
 - Modeling of multiple types of stutter (+1, -1, -2, +0.5, -0.5), based on the longest uninterrupted sequence of the short tandem repeat (STR) motif
 - Shoulder peaks, determined by the size/position of a peak relative to an allele peak
 - Peak height ratio expectations based on the amount of DNA
 - Degradation, drop-in, and dropout probabilities, based on the amount of DNA

• LR calculations and robustness analysis:

- Build scenarios from simple to complex, with assumed contributors or multiple potential contributors
- Test the significance of your results through simulations with the robustness algorithm

Profile review

Start your interpretation with a big-picture view of your entire project. Manage, evaluate, and compare multiple samples at once. In the Profiles screen (Figure 1), you will find genotypes, sample plots, sample quality information, and NoC estimates.



Figure 1. Manage, review, and edit sample information across a project in the Profile screen where sample quality, electropherograms, and analysis details are displayed (e.g., degradation, height distribution, analysis performed).

NoC estimation

Multiple algorithms are available within GeneMapper *PG* Software to estimate the NoCs to a mixture, which is a critical step of the interpretation process. From the simplest methods like MAC and total peak analysis (Figure 2) to machine learning–based methods such as decision trees (Figure 3), you can examine the estimation process with full transparency. No matter which method you choose, each is fully traceable and easy to explain.





Figure 2. NoC estimation using the total peaks algorithm. The total peaks algorithm computes peak statistics on a set of simulated samples, then shows the resulting value for the currently selected samples.

Figure 3. The NoC decision tree with inputs related to the DNA profile: the number of peaks within the profile, minimum and maximum number of alleles at a locus, and allele heights.

Fully continuous probabilistic model

GeneMapper *PG* Software models DNA profiles at the global level, applying consistent contributor proportions across loci for biologically grounded results. The system integrates detailed models for stutter artifacts, shoulder peaks, dropout, drop-in, baseline noise, and peak height ratios, tailored to DNA quantity, amplification conditions, and forensic STR kits. For the mixture deconvolution step, GeneMapper *PG* Software employs a structured, systematic approach following a set of rules executed in fixed order through the deterministic BCOS algorithm, helping to deliver efficiency and precision. With this approach, results remain the same for any given set of parameters.

Explore analyses in detail with step-by-step explanations and robust visuals. Instantly view electropherograms comparing observed DNA profiles to expected solutions, extract major contributors, export CODIS-compatible files for database searches, and search deconvoluted profiles against other samples in your project—all in one place (Figure 4).



Figure 4. Deconvolution analysis: easily view profiles, electropherograms, fit, and weight all in one place.

Compare profiles

GeneMapper *PG* Software provides powerful tools for comparing DNA profiles using algorithms across several intuitive screens. The Heat Map screen allows users to calculate and visualize matches between two sets of profiles, while the Match Details screen offers detailed comparisons of individual profiles. The Match Table screen presents a list of the potential matches filtered by percentage, making it easy to identify the most relevant profiles. Whether for contamination detection or examining pattern cases, these features streamline the comparison process, enhancing the ability to analyze and interpret complex DNA data efficiently.

LR calculations

Calculate even the most complex LR scenarios with an intuitive interface (Figure 5) where you can easily build the scenario, visualize the result, and drill down into the calculation details. The LR Robustness analysis tool simulates thousands of profiles, visualized through an empirical cumulative distribution plot, helping you determine whether your LR result is significant or the result of a chance match (Figure 6). The simulated profiles can be configured to be unrelated to the person of interest, or a specific family relationship.





Hispanic



Figure 6. Robustness. Plot your POIs LR against likelihood ratios of simulated (unrelated or related) profiles to determine weight of association.

Transparency

Ultimately, it's not just about getting an answer—it's about interpreting the data with clarity and communicating your findings with ease. The GeneMapper *PG* Software's deterministic algorithm, step-by-step explanations, and powerful visualization tools help make analysis clear and easy to understand. Embrace the next phase of forensic genotyping with GeneMapper *PG* Software and experience the difference in every detail.

Supported file types

GeneMapper *PG* Software v1.0 offers robust support for multiple file types, facilitating easy integration with a variety of data sources. Compatible file formats include SER project files from Applied Biosystems[™] GeneMapper[™] *ID-X* Software and CODIS CMF 3.3 files, as well as CSV and TXT files from various applications, including GeneMapper *ID-X* Software. To facilitate accurate analysis, files must minimally contain sample names, marker names, allele designations, calls, and peak heights. This versatility allows for efficient and comprehensive data management, enhancing the overall workflow and helping to ensure compatibility with existing systems.

Validation

Thermo Fisher Scientific has validated GeneMapper *PG* Software v1.0 following the FBI Quality Assurance Standards (July 1, 2025) and guidelines from the Scientific Working Group on DNA Analysis Methods (July 1, 2025). Each laboratory using GeneMapper *PG* Software v1.0 should perform its own internal verification or validation studies to establish interpretation criteria and demonstrate that the software is appropriate and fit for its own HID uses.

Over the years, advancements in DNA extraction, STR chemistry, and capillary electrophoresis (CE) instrumentation have expanded the amount of DNA that can be recovered from more samples than ever before. With increased sensitivity and the high demand for DNA evidence, mixture interpretation remains one of the foremost challenges. GeneMapper *PG* Software extends the utility of widely adopted GeneMapper *ID-X* Software with an intuitive suite of software designed to bring clarity, deeper insight, and confidence to your mixture analysis.

To learn more, refer to the GeneMapper *PG* Software v1.0 Getting Started Guide (Pub. No. MAN1000548).

Computer specifications

The recommended supported computer hardware and system requirements for GeneMapper *PG* Software are as follows:

- Minimum specifications:
 - Processor: 12th gen Intel Core[™] processor or equivalent with 10 or more cores
 - Installed RAM: 16 GB (15.6 GB usable)
- Recommended specifications:
 - Processor: 11th gen Intel Core[™] processor or equivalent with 10 or more cores
 - Installed RAM: 32.0 GB (31.4 GB usable)

Supported operating systems

Microsoft[™] Windows[™] 10 or higher, 64-bit OS

Ordering information

Description	Cat. No.
GeneMapper PG Software v1.0	A77500

Visit <u>thermofisher.com/gmpg</u> or contact your local sales representative to find out more about the software.

Learn more at thermofisher.com/gmpg

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