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GeneMarker® : DEVELOPMENT AND VALIDATION OF A NEW SOFTWARE TOOL FOR THE ANALYSIS OF STR DATA

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The analysis of STR data generated from forensic evidence typically requires specially designed software. Essential features of the analysis software include: 1) A user-friendly interface and functionalities, 2) A multitude of strategically selected user-defined parameters, 3) Accurate processing of raw data, 4) Quality assessment of analyzed data, 5) A mechanism for flagging those profiles that should be looked at first when doing technical review, 6) Effective management of data and associated files, and 7) Adaptable reporting features. For the analysis of data generated using Applied Biosystems instrumentation (e.g., the 310 and 3100 Genetic Analyzers), the only analysis software readily available and forensically validated is Genotyper® and GeneMapper® ID, with GeneMapper® ID being the only software package that provides quality scoring and flagging of potentially poor profiles. Through a research-based collaboration with the Forensic Science program at Penn State University and SoftGenetics®, a company located in State College, Pennsylvania, a forensic version of the software package GeneMarker® was developed. The forensic version of GeneMarker® meets or exceeds the criteria listed above and is an attractive alternative to GeneMapper® ID for laboratories doing forensic STR analysis.

GeneMarker®, which utilizes advanced sizing algorithms, takes the raw data files generated on any Applied Biosystems instrument and analyzes the data in accordance with forensic interests and principles (i.e., GeneMarker® complies with the format of forensic STR multiplexes and associated allelic designations). The analyzed data is presented in a user-friendly manner and provides functionalities that meet or exceed the interests of the forensic community. During the development of the forensic version of GeneMarker®, SoftGenetics® focused on key areas of the analysis process: e.g., data management, data analysis, and reporting. Once the software had been modified to address the desired features and functions, validation studies were performed by doing side-by-side analysis of STR profiles from single source, mixed, and LCN samples, comparing the data analyzed with GeneMarker® with data analyzed using an existing forensic STR analysis software package. In addition, user-testing was conducted on actual forensic STR data in public and private forensic DNA laboratories. The figure below is an illustration of STR data at the D8S1179 locus using the Identifier® multiplex and analyzed with GeneMarker®.

SoftGenetics® has been recognized by Applied BioSystems as a valued 3rd party provider of analytical software for their electrophoresis systems.

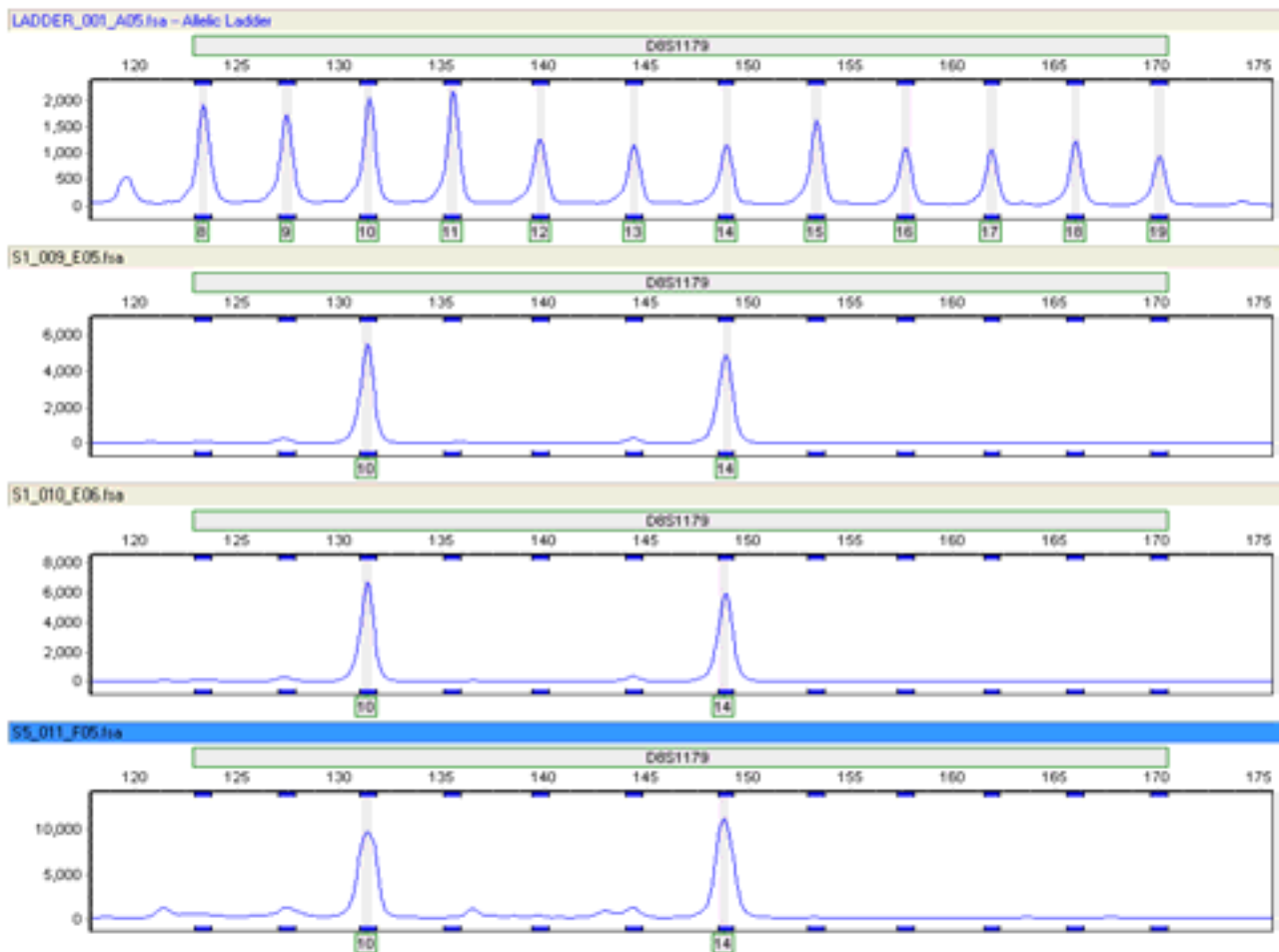


Figure: Identifier data at the D8S1179 locus for DNA extracted from a saliva sample and amplified using different quantities of DNA (approximately 1, 5, and 10 ngs, top to bottom)