AN ASSESSMENT OF THE GENEMAPPER® ID-X MIXTURE ANALYSIS SOFTWARE TOOL FOR THE INTERPRETATION OF MIXTURES WITH TWO CONTRIBUTORS

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The development of expert systems to aid scientists in the analysis of forensic DNA profiles is of great interest to the forensic community. Of particular appeal are computer software programs that can analyze DNA results with the same proficiency as manual interpretation conducted by an experienced analyst. GeneMapper® ID-X (GMID-X, v1.3) is a software package developed as an expert system for single source samples, and includes a mixture interpretation tool to minimize the amount of time analysts spend on data interpretation. This latter element of the software was used to interpret two-person mixtures in a controlled study. Eight different DNA samples, provided by NIST, were selected with a minimum number of shared alleles and a maximum number of heterozygote loci. From these samples, five sets of mixtures were prepared in ratios of 1:1, 1:2, 1:4, 1:6, 1:8, 1:10, 2:1, 4:1, 6:1, 8:1, and 10:1, and amplified using the Identifiler® and Identifiler® Plus kits following the recommended protocols. The GMID-X mixture analysis and deconvolution feature was compared to manual interpretation.

The mixture analysis module of GMID-X was superior to manual analysis when pulling out major and minor component profiles for the samples associated with 1:2, 2:1, 1:4, and 4:1 mixture ratios. It is generally difficult to determine which alleles should be in the major and minor components when doing manual analysis, but the software was able to do this correctly on a routine basis. Conversely, and somewhat surprisingly, manual analysis provided better deconvolution of the 1:10, 10:1, 8:1, and 1:8 mixtures. For some samples at these ratios, the software had a hard time determining the minor components, especially at 4 allele loci. Occasional dropouts were selected even when the alleles at the locus were above the stochastic threshold (ST) used in the manual interpretation. In a few instances, the alleles at a locus were below the ST, but no dropouts were considered. Some alleles were not chosen to be part of the minor component when they were clearly present, due to the software not utilizing a ST for the analysis. Instead, a mixture interpretation threshold was used as a global cutoff filter, meaning that alleles with heights below 150 RFU (minimum peak height used for analysis in the current study) were not considered. This also caused problems with the minor component being pulled out properly. In order to overcome this issue, analysts can lower the mixture interpretation threshold (MIT) and reanalyze the data. Occasionally, there were problems with the genotype selection; for example, no major component genotype was selected, the genotypes selected had high residual values, and/or the unselected genotypes had lower (better) residual values. This was attributed to peak height ratios not meeting the threshold.

Overall, the mixture analysis software tool within GMID-X (v1.3) was found to be easy to use and has the potential to make the mixture interpretation process faster and easier for forensic DNA analysts; remembering that GMID-X (v1.3) is not an expert system for mixture analysis. ⌘