

IMPLEMENTATION OF THE AB 3130 GENETIC ANALYZER AND GENEMAPPER® ID V3.2 IN FORENSIC CASEWORK

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Molecular techniques used to generate and analyze genetic data require significant user intervention and technical review. However, the introduction of GeneMapper® ID analysis software, developed by Applied Biosystems, has enabled the forensic community to streamline current processes and ensure timeliness without any reduction in the quality of the data. The Centre of Forensic Sciences (CFS) has recently completed validation of the AB 3130 Genetic Analyzer and GeneMapper® ID v3.2 analysis software for use in a high volume work stream. Validation, in accordance with current SWGDAM guidelines, is outlined here. Initially, GeneMapper® ID v3.2 was only validated using the basic software functions, however; additional data is presented which illustrates the full functionality of the software. Specifically, the use of Process Quality Values (PQVs), which quickly identify genotype quality, is demonstrated on both mixtures and single-source samples, typically observed in casework. Three injection parameters were validated for use on the AB 3130: 1kv for 15 seconds, 1kv for 20 seconds, and 1kv for 25 seconds. Studies demonstrated that sample-to-sample carry over does not occur between injections on the AB 3130. Sizing precision, determined through multiple injections of AmpFISTR® Profiler Plus™ and CoFiler™ allelic ladders, as well as Promega's PowerPlex® Y and PowerPlex® 16 allelic ladders, was within the 0.5 base pairs window for all alleles. Sensitivity studies demonstrated that full profiles were obtainable with as little as 60pg of template DNA, although peak height discordance was noted. The ability of the instrument to detect the minor component in mixtures was also investigated. DNA from two individuals was combined such that the ratio of the major to minor component was 1:1, 5:1, 10:1, 15:1 and 20:1. All minor alleles were detected in mixtures of up to 20:1 using the 1kv, 25-second injection parameter. Shorter injection times resulted in two minor alleles falling below the threshold value of 50 relative fluorescent units (rfu). Resolution studies demonstrated that single base pair microvariants were resolved in mixtures ranging from 5:1 to 15:1 depending on locus. Finally, profiles generated using NIST traceable reference materials and other non-probative forensic samples were concordant with those previously analyzed on either the ABI PRISM® 377 or 3100 platforms using GeneScan® and Genotyper® software packages currently operating within the CFS. Our validation studies have demonstrated that the 3130 Genetic Analyzer in combination with GeneMapper® ID is a reliable and robust platform suitable for casework. Furthermore, the dual functionality of GeneScan and Genotyper as well as the use of PQVs in GeneMapper® ID reduces the amount of time required for data analysis by 3 hours, per 96-well plate.