VALIDATION OF GENEMAPPER® *ID-X* SOFTWARE V1.0.1 EXPERT SYSTEM WITH AMPF/STR® IDENTIFILER® DATA

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The CAL-DNA Data Bank Program hosts the fourth largest offender DNA database in the world. The number of samples the CAL-DNA Data Bank Program (Data Bank) receives for processing dramatically increased with the passage of Proposition 69 in California in 2004 that immediately qualified all felony offenders and required collection from all adult felony arrestees starting in January 2009. Most of the Data Bank process already uses automation for liquid or sample handling. However, data analysis and review have remained more labor intensive, and are potential bottlenecks in sample processing. We conducted a validation for NDIS approval of the GeneMapper® *ID-X* Software v1.0.1 Expert System (GMID-X), using the ABI 3130xl Genetic Analyzer and Identifiler®, to alleviate this bottleneck.

This poster presentation describes the validation of GMID-X as an expert system for use at the CAL-DNA Data Bank Program and the impact of its use on STR data interpretation throughput. Prior to the GMID-X validation, GeneMapper® ID v3.2.1 (GMID) was utilized at the Data Bank for data analysis. All samples, controls, ladders and LIZ size standards were manually reviewed by the analyst using GMID. This manual review requirement was largely responsible for the analysis bottleneck. In the first six months of 2009, however, due to adult felony arrestee collections, monthly submissions to Data Bank doubled over that of the previous six months. Using an expert system for data analysis, the Data Bank planned to reduce analysis time and, therefore, eliminate the backlogs resulting from increased submissions. The poster provides a model for other laboratories conducting expert system validations, internally or for NDIS acceptance, and describes how such an expert system assists a laboratory processing high volumes of DNA profiles.

For such an expert system to be worthwhile, all samples flagged as acceptable must also pass according to the laboratory's standards. To test the software and ensure that all passing samples were of sufficient quality for upload to the Combined DNA Index System (CODIS), specific parameters were established for GMID-X. Selected sample sets with known problems were analyzed with GMID-X in a calibration study (involving 426 samples) that provided the data required to set analytical and interpretational thresholds. These thresholds and parameters were adjusted so that any anomalous data peaks would be flagged by the software for manual review. Beyond the usual fine-tuning, marker range sizes and bins were adjusted at some loci so that ambiguous alleles falling between two loci would be flagged for review. Use of size range filters also reduced the number of reproducible artifact peaks flagged. In addition, the GMID-X provides some additional quality parameters, not provided with GMID, for evaluating both LIZ size standards and allelic ladders. After evaluating the effects of implementing these parameters, settings were identified that eliminated manual review of passing allelic ladder and LIZ samples and significantly reduced analysis time.

After establishing "Analysis Method, Panels and Bins" parameters in GMID-X, concordance studies were performed using data from 1479 samples. All samples were typed with GMID-X, as well as GMID. Genotype tables for all samples were compared for concordance. Any sample that passed utilizing the GMID-X parameters was manually reviewed to ensure that the profile met Data Bank standards for CODIS. Excluding the calibration samples selected for the study, GMID-X analysis of typical Data Bank data sets resulted in a 66.8% pass rate, that is, an average of 33.2% of samples per representative data set would need review (vs. 100% manual review with GMID).

The poster demonstrates that in our Laboratory, analysis of single source DNA samples using GMID-X according to our protocol produces results that are "as good as or better" than using the current Data Bank GMID manual review protocol. When GeneMapper *ID-X* is used as described, this validation demonstrates that it can be implemented as an Expert System where "passing" sample profiles do not need additional manual review. The time required to successfully analyze a plate can therefore be greatly reduced when compared with the current GMID "manual review" processes.