B100 Battelle's Final Report on the National Institute of Justice (NIJ) -Sponsored Initiative: A Feasibility and Guidance Study of Massively Parallel Sequencing (MPS) for Forensic DNA Applications

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After attending this presentation, attendees will better understand the MPS technology, its potential viability for forensic applications, and the identified gaps and considerations for implementation perspectives.

This presentation will impact the forensic science community by providing an awareness of MPS technology advancements in the field of forensic DNA analysis, the potential strengths and limitations of the specific technology application, and critical information relevant to the laboratory-specific decision process for potential investment in such a technology.

Forensic DNA analysis through Capillary Electrophoresis (CE) -based typing of Short Tandem Repeat (STR) is a well-established and successful technology with widespread legal and technical acceptance and is integral for the generation of more than 16 million DNA profiles registered within the Federal Bureau of Investigation's (FBI's) National DNA Index System (NDIS). The emergence of MPS presents opportunities for potential success beyond conventional CE-based techniques, specifically with respect to degraded specimens (missing persons) and possibly complex samples (mixtures). More significantly, MPS provides a broader scope of informative and discriminating data through Single Nucleotide Polymorphisms (SNPs) for identity, physical appearance, ancestry, and kindred relationships. Consistent with newer technologies, MPS also introduces levels of change, some of which are disruptive to the present approaches routinely applied by forensic DNA laboratories, ranging from nomenclature, workflow, and instrumentation, to data interpretation and reporting.

In 2014, Battelle first reported the receipt of a National Institute of Justice (NIJ) grant for initiating a two-year study (2015-17) to assess the technical readiness and feasibility of the MPS technology for forensic applications. The study consisted of both Performance Testing (Phase 1) to assess system capabilities and optimization of commercially available MPS products, and Inter-Laboratory Testing (Phase 2) for conducting a series of carefully designed studies to address key validation criteria. The Phase 2 effort specifically included the integral testing participation of technical leads from the Armed Forces DNA Identification Laboratory (AFDIL); the Bureau of Alcohol, Tobacco, Firearms and Explosives (ATFE); the California Department of Justice (CAL DOJ); the FBI; Harris County Institute of Forensic Sciences (HCIFS); the National Institute of Standards and Technology (NIST), the New York City Office of the Chief Medical Examiner (NYC OCME), and the North Carolina State University. The validation study included assessment of the technology with respect to reproducibility, precision, concordance, sensitivity, mixtures, and non-probative casework. For the latter, each participating laboratory selected up to 14 samples, varying in types and often reflective of the respective missions for each laboratory. This presentation will convey the final results of this comprehensive study, including strengths, gaps, and considerations of strategic roadmaps for the MPS technology acceptance and transition. In total, a contemporary assessment of the technical readiness of the MPS technology will be provided, including examples of actual casework processed across two of the commercially available analysis workflows.

Massively Parallel Sequencing, Abundance, Iso-Allele

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