Battelle's Final Report on the NIJ Sponsored Initiative:
Feasibility and Guidance Study of Massively Parallel Sequencing for Forensic DNA Applications

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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 integral for the generation of over 16 million DNA profiles registered within the FBI's National DNA Index System (NDIS). The emergence of Massively Parallel Sequencing (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 (SNP's) 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 an NIJ grant (Award Number 2014-DN-BX-K023) for initiating a two-year study (2015 - 2017), 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 Federal Bureau of Investigation (FBI); Harris County Institute of Forensic Sciences (HCIFS); the National Institute of Standards and Technology (NIST), the New York Office of the Chief Medical Examiner (NYOCME) 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 to include 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 massively parallel sequencing technology will be provided, to include examples of actual casework processed across two of the commercially available analysis workflows.

A final report has been submitted to National Institute of Justice and separate manuscripts, further detailing key components of this project, are presently under development.